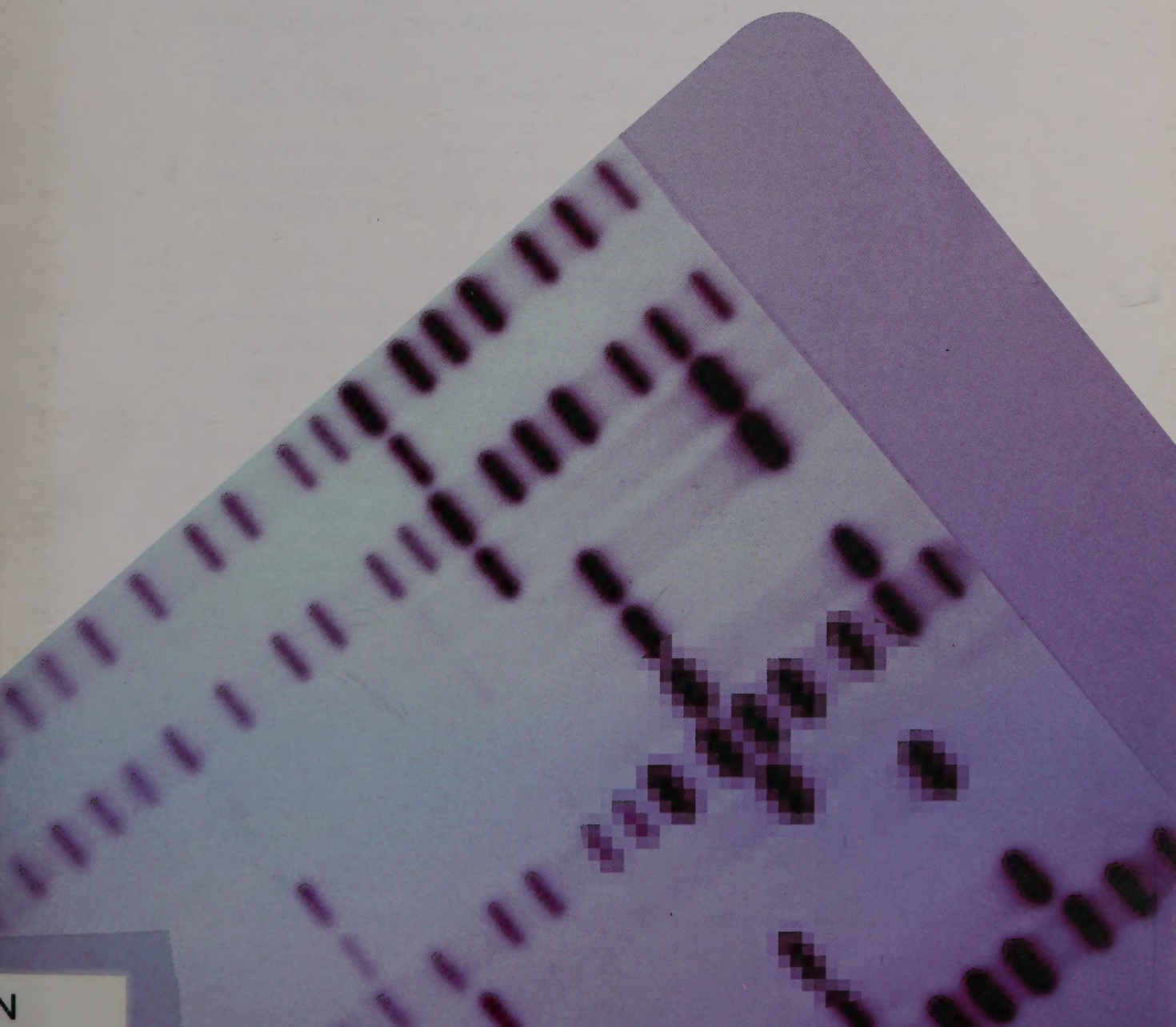




Human
Genetics
Commission

Whose hands on your genes?

A discussion document on the storage protection
and use of personal genetic information



Whose Hands On Your Genes?

A Summary

Please tick the boxes that apply ☒ and use a separate sheet if you would like to add further information. This form can be sent to the Human Genetics Commission by freepost, see page vi.

A. Personal genetic information

(This is summarised inside the front cover, more information is given in sections 2 and 3 of the main document.)

What is genetic information?

- We have been using a person's family history to get genetic information for a long time. Developments in the techniques for analysing DNA from bodily samples mean that we have new and more accurate sources of genetic information available to us. These open up new possibilities and challenges. In this document the HGC is mainly concerned with genetic information obtained from these new techniques. Some people have suggested that this type of genetic information should be considered as different to the information that comes from family history and external examination.
- Is genetic information special? The law protects the confidentiality of personal information. Genetic information is a type of personal information and perhaps should therefore be treated like any other medical information about a person, such as information in their GP records. But there are several reasons why genetic information may be seen as special and should be treated differently:
 - it is unique to each person (except for 'identical' twins);
 - it is technically possible to obtain it from a very small bodily sample without a person's consent;
 - it can be used to predict disorders a person may develop in the future and may tell other family relatives about disorders they may develop too;
 - these predictions can be of interest to others such as insurance companies or employers; and
 - it has a potential commercial value to organisations introducing new developments based on genetic information.
- Others may feel that the current rules are enough to protect genetic information. It could be treated the same way as other medical information, like the results of a test for diabetes or HIV, and may not need special recommendations.

Personal genetic information

A 1) Is HGC right to focus mainly on information made available by new DNA analysis techniques rather than on all genetic information such as that based on family history?

Yes ☐ No ☐

A 2) Does the confidentiality of genetic information need to be protected in a different way to other personal medical information?

Yes ☐ No ☐ It depends ☐

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WHOSE HANDS ON YOUR GENES?

A discussion document on the storage, protection and use of personal genetic information

The Human Genetics Commission (HGC) is an independent body that advises the Government on policy issues in the field of human genetics. You can find out more about us on our website at www.hgc.gov.uk.

We are currently looking at the uses of personal genetic information, to see what if anything should be done to address people's concerns about possible abuses of such information. We have identified a broad range of questions about these issues which we are putting to public consultation, and will then draw up our recommendations in 2001.

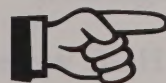
These questions are set out in detail in the main part of this document, along with relevant background information. We have also summarised key issues and questions in the pull out section inside the front cover, for people who want to give a shorter response.

HGC welcomes responses to any or all of the questions in both the main part of the document and the summary by 28 February 2001.

We may wish, in future, to make the responses to this consultation exercise publicly available. If you are happy with this, please tick the appropriate box, otherwise your views will be treated in confidence.



Summary and
tick box responses



Full discussion document
and detailed responses

Please let us have your comments on the way this consultation exercise has been carried out, there is an evaluation form in the main document on page 47.

THANK YOU FOR TAKING THE TIME TO READ THIS DOCUMENT

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B. Why people have genetic tests

(This is summarised inside the front cover, more information is given in sections 5, 6 and 7 of the main document.)

Confidentiality of genetic information from tests for medical reasons

- Giving **consent** for tests for medical reasons: the law says that, except in special circumstances, a patient should not be treated without his or her consent. People need a certain level of information about a treatment and its consequences so that they understand what they are consenting to.
- **Confidentiality**: the results of a genetic test are personal. But they can also have important implications for other family members. In most cases people taking genetic tests agree that other family members should be told about any implications for them. But sometimes people may not want to pass such information on. In these cases a third party, eg a doctor, may feel the situation is serious enough that they should tell family members who may be affected. This would be a breach of the person's confidence.

Confidentiality of genetic information from tests for medical reasons

B 1) Should details of a person's genetic test results ever be passed on to other family members without that person's consent?

Yes ☐ No ☐ It depends ☐

Genetics and medical research

- Researchers find medical databases of genetic information and collections of stored tissue samples invaluable in helping them understand and cure human diseases.
- Some research would benefit from the study of new genetic information from samples that have been taken from patients for other reasons, eg blood tests or tumours after surgery. These samples could be used either anonymously or in a way that identifies people but it may not be possible to get people's consent to use such samples. Using these samples may lead to important advances in curing disease beyond those first thought of at the beginning of a study.
- As for all medical research, it is important that people give their consent for their genetic information to be put on such a database. If a person agrees to give a sample, some feel that he or she has agreed to research in general, and the sample can be used in a range of studies. Others feel that the person should be asked to give their consent each time their sample is used for a new study. There are several possible types of study – sometimes the samples will be anonymous, sometimes they will be coded (so the researchers can't identify the people who gave samples) and sometimes the people involved will be identified.

Genetics and medical research

B 2) Should a person's genetic information be put on a medical research database without their consent?

Yes ☐ No ☐ It depends ☐

B 3) Should people be asked to give consent each time new research is carried out using their sample?

Yes ☐ No ☐ It depends ☐

What is personal genetic information? (section A)

- **Genetic information** is found in almost every cell in our bodies. Most cells contain 23 pairs of **chromosomes** – one of each pair inherited from each parent. Chromosomes are made of long chains of **DNA** and individual segments of the DNA are known as genes. Genes carry instructions that influence particular characteristics eg eye colour or blood group. It is the **differences** between their **genes** that make people different.
- Sometimes a single gene can affect the chance of developing a disease eg cystic fibrosis. More often, a number of genes and a person's lifestyle and environment will all play a part. Because of this a person's genetic information alone won't tell us what diseases they might get in the future.
- Other segments of DNA between genes can be used to identify people or see if they are related to each other.

How do we get genetic information?

- **Family history:** we know that, for some rare inherited diseases, if someone has that disease their child has a 50% chance of inheriting the disease and their grandchild has a 25% chance.
- **External examination:** we can find out some things about a person's genetic makeup by looking at their appearance (eg eye colour).
- **Analysis of a bodily sample:** DNA can be analysed from a sample of a person's cells. These cells can be taken from nearly any bodily tissue: a mouth swab, a few hairs or flakes of skin or a spot of blood.

Why people have genetic tests (section B)

- **Genetic tests for medical reasons:** tests can help to identify and treat a disease. They can also tell a person whether or not they are likely to develop diseases. And they can tell a person with a family history of a disease if they are likely to pass this on to their children.
- **Genetic testing for research reasons:** tests may also be undertaken by researchers to expand our understanding of genetics and disease.

Uses of genetic information (section C)

- Other organisations may be more and more interested in a person's genetic test results in the future. For example, insurance companies could use this sort of information when they set a person's insurance premiums. Or employers could use it to tell them more about potential employees.
- Genetic information can also be used for forensic reasons, to identify or eliminate a suspect from police enquiries. This information is stored on the National DNA database for future use in crime detection.
- Genetic information can be used to tell if someone is a child's parent. The information can help settle disputes about child maintenance payment or for immigration reasons.

B 4) Should commercial medical researchers have access to these databases?

Yes ☐ No ☐ It depends ☐

B 5) Should samples taken for other reasons be used for research without the consent of the people who gave them:

(1) if the samples are anonymous Yes ☐ No ☐ It depends ☐

(2) if the samples can be traced to a person Yes ☐ No ☐ It depends ☐

C. Uses of Genetic Information

(This is summarised inside the front cover, more information is given in sections 8 and 9 of the main document.)

Genetics and insurance

- Insurance companies usually ask for as much medical information as possible when assessing an application for life or health insurance. A person applying for insurance may be asked about their family medical background and lifestyle. They may be asked to take a medical examination. Under a voluntary code of practice, insurance companies in the UK will not ask people to take a genetic test. But if the person has already taken a test the companies may sometimes ask for details of the results.
- Some people feel that insurance companies may unfairly discriminate against those whose genetic tests show they are at greater risk of developing certain conditions. It may mean some people are not able to get insurance. Insurers have argued that genetic information is no different from any other medical information in this context. They feel they should be allowed to use the results of genetic tests to help them assess the nature of the risk and the level of insurance premiums. Some say that other policyholders may pay higher premiums if insurers can't see the results of genetic tests.

Genetics and insurance

C 1) Should an insurance company be able to ask to see any results of a person's genetic tests to assess whether their premiums should go up or down?

Yes ☐ No ☐

C 2) If yes to the above, should there be controls on the way the information can be used?

Yes ☐ No ☐

C 3) If yes to the above, should any of the following affect whether or not the information should be used:

(1) the type of insurance (life/health etc)? Yes ☐ No ☐

(2) the type of genetic condition? Yes ☐ No ☐

(3) the value of the policy to be insured? Yes ☐ No ☐




Tuck A into B to form envelope
Please do not staple or glue

----- First Fold -----

Baroness Helena Kennedy
Chair, Human Genetics Commission
Freepost LON15502
Maidenhead
SL6 2BZ

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 We would be grateful if you could provide us with your contact details to aid in our analysis of responses and to let us know if you wish to receive further information about this or future HGC consultations.

(BLOCK CAPITALS PLEASE)

Title _____ Forename/s _____ Surname _____

Organisation (if applicable) _____

Address _____

Postcode _____ e-mail _____

I would be happy if my response were made publicly available ☐ I would be interested in future consultations ☐ I would like a summary of the outcome of this consultation ☐



Genetics and employment

- In the future some **employers** might want to use genetic tests for several reasons, eg: to identify employees who may put their colleagues or members of the public at risk in the workplace; who may be more likely to get an occupational disease; or who are likely, because of genetic factors, to experience long periods of absence from work. There has been some public anxiety that employers will use genetic tests to discriminate unfairly against employees who are seen to be at risk of a particular illness or condition.
- At the moment there is no law stopping employers from using the results of genetic tests. An employer could require someone to take a genetic test before deciding whether or not to offer him or her a job. There is no evidence that this is being done in the UK (when the issue was examined in detail, only one employer was found to be using genetic testing). Other countries do have laws in this area – either to ban employers from using tests, or only allow their use if there is a clear health requirement for the job, or to protect an employee's health in the workplace, or to protect employees against genetic discrimination.

Genetics and employment

C 4) Should an employer see the results of employees' genetic tests:

- (1) to see if employees may become a risk to colleagues
or members of the public in their work?

Yes ☐ No ☐ It depends ☐

- (2) to see if employees are likely to become prone to an inherited
disease or disability?

Yes ☐ No ☐ It depends ☐

- (3) to see if they may be sensitive to certain substances that they will come
in to contact with in their job?

Yes ☐ No ☐ It depends ☐

Genetics and police databases

- The police can take a bodily sample from anyone suspected of a wide range of offences. The DNA profile obtained from the sample can then be compared with other profiles from evidence taken from crime scenes. These DNA profiles are stored on a national database to help identify or eliminate suspects in the future. DNA forensic databases have been widely recognised as being as important as fingerprint records in crime detection. But genetic information is different as it tells you more about a person than a fingerprint does. The DNA profile can only be used to identify a person and will not (as far as is known) provide any information about genetic disorders. It would be possible to use the sample itself to provide such information at a later date.
- There is public support for the forensic use of genetic information. But some concerns have been expressed about certain aspects of the existing arrangements, such as when the police should take a sample and how that sample is kept.

Genetics and police databases

C 5) Should the police take bodily samples from people suspected of:

- | | | |
|----------------------|------------------------------|-----------------------------|
| (1) burglary? | Yes <input type="checkbox"/> | No <input type="checkbox"/> |
| (2) drink driving? | Yes <input type="checkbox"/> | No <input type="checkbox"/> |
| (3) murder? | Yes <input type="checkbox"/> | No <input type="checkbox"/> |
| (4) sexual offences? | Yes <input type="checkbox"/> | No <input type="checkbox"/> |
| (5) shoplifting? | Yes <input type="checkbox"/> | No <input type="checkbox"/> |

C 6) If someone is charged with a crime, and his/her DNA profile is placed on a police genetic database but they are later acquitted of the crime, should the sample be taken off the police genetic database?

Yes ☐ No ☐

C 7) Should the police have access to information on other genetic databases, eg medical databases, to cross check against the forensic database?

Yes ☐ No ☐

Genetics and family relationships

- The segments of DNA between genes vary a lot between unrelated people but are very similar between blood relatives. This means they can be used to see if people are related to each other. Most commonly this is used to confirm paternity. The results of paternity tests can be used in deciding child maintenance or inheritance disputes. They can also be used for immigration reasons.
- Until recently blood samples were needed from the child and both parents to do these tests. New techniques allow tests on hair or mouth swab samples. These techniques mean that only the possible father and the child now need to give samples, and a test could be done without the mother knowing it. They have also led to sampling kits being advertised via the Internet and sent through the post. These developments have implications for consent to testing.

Genetics and family relationships

C 8) Should tests for family relationships be carried out without the consent of all the people involved?

Yes ☐ No ☐ It depends ☐

Thank you for taking the time to complete this questionnaire

Please send your comments to:

Baroness Helena Kennedy
Chair, Human Genetics Commission
FREEPOST LON15502
Maidenhead
SL6 2BZ

Further copies of this document only are available by writing to:

'Whose hands on your genes?'

PO Box 777,
LONDON SE1 6XH

or by faxing:
01623 724524

or by emailing:
doh@prolog.co.uk

or via the HGC website at www.hgc.gov.uk

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1. Introduction

- 1.1 Progress in molecular biology means that we know an increasing amount about our genetic makeup. This information may be general, in the sense that it is about our common genetic heritage, the human genome – the genes we all share and how these may function. Or it may be very specific information about genetic variations that are more or less unique to an individual. This latter information is clearly very personal, and just as there have been expressions of welcome for progress in genetic science and all that it means for medicine, there have at the same time been strong concerns about privacy and possible genetic discrimination.
- 1.2 The Human Genetics Commission (HGC), which has the task of advising Government on policy issues in this area, has established a working group which is currently examining how personal genetic information is treated and identifying what may need to be done to ensure that concerns over the potential abuse of such information are addressed. The terms of reference and membership of the Working Group are given in Annex 1 and a full list of HGC Members is given in Annex 2.
- 1.3 The Working Group has now identified a broad range of questions which HGC wishes to put to public consultation before drawing up recommendations in 2001. These questions are set out in this document, along with a brief explanation of the background in each case. The aim of the consultation is to ascertain the views of members of the public and of interested organisations in the UK on these issues. This is intended to be a preliminary document to identify the key issues in these areas that will inform HGC in the development of its final report and recommendations to Ministers.

The consultation process:

November 2000 ⇒ February 2001: this consultation on the key issues;

March ⇒ April 2001: HGC to consider responses and gather additional material;

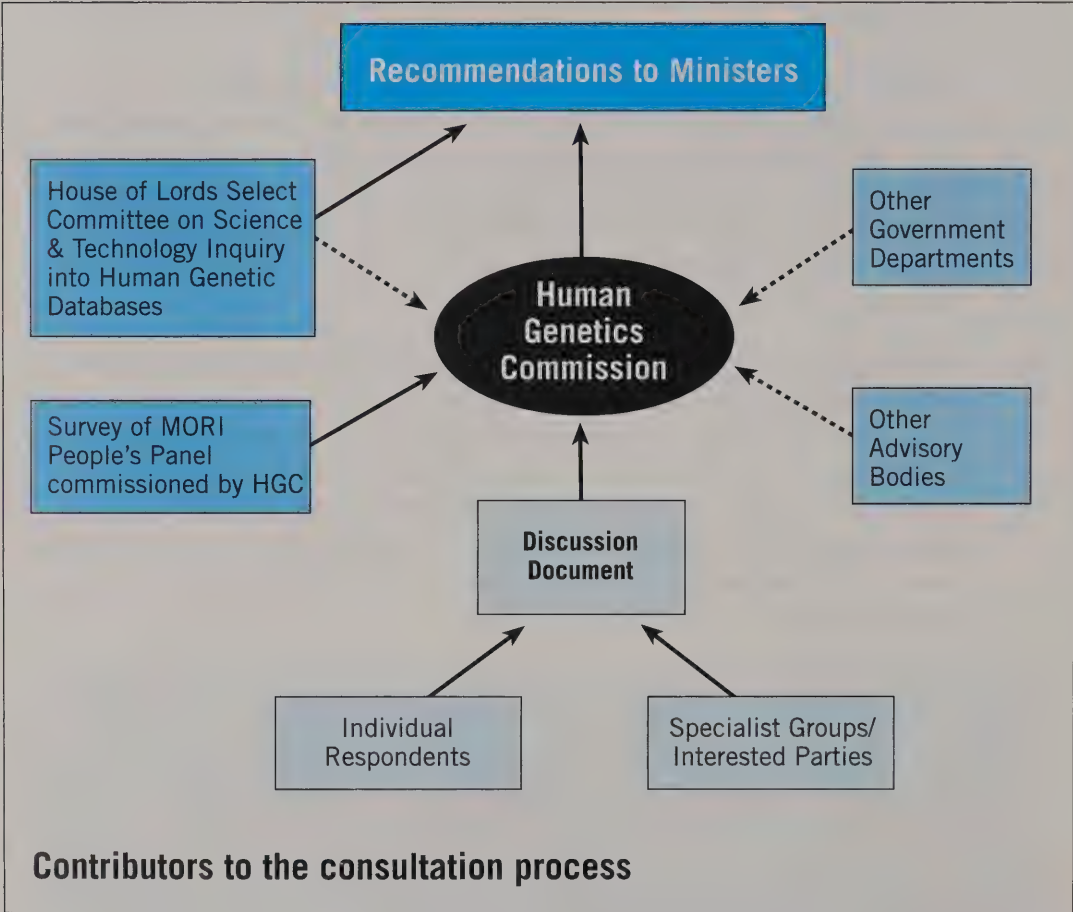
March ⇒ June 2001: prepare and consult on draft report;

Late 2001: Report to Ministers.

House of Lords Inquiry into human genetic databases

- 1.4 There is also an Inquiry under way by the House of Lords Select Committee on Science and Technology investigating current and planned activities in the operation of human genetic databases, and the opportunities and concerns arising. The Select Committee and the HGC liased to ensure that there was minimum overlap between the two exercises and, indeed, to provide appropriate complementarity. Given the Working Group's remit, the Select Committee is investigating neither public attitudes towards such databases nor what the regulatory structures should be. However, its findings (which are due to be published at around the end of March 2001) will doubtless bear on these matters and be of use to both the HGC in framing its advice to the Government and to the Government in considering that advice.

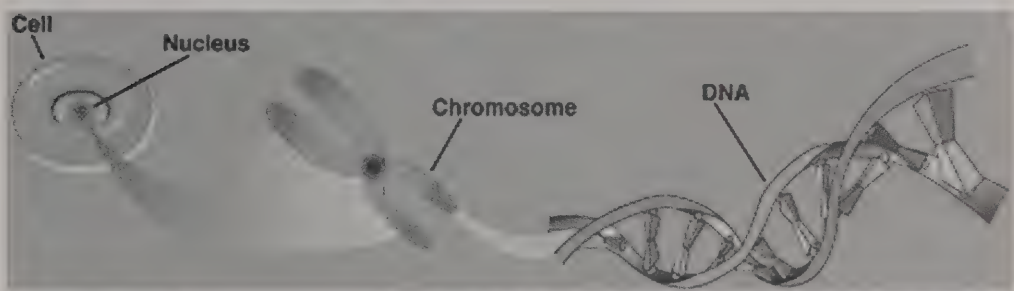
- 1.5 The Select Committee issued its call for evidence in July 2000, seeking responses principally from the bodies involved in maintaining, developing or using human genetic databases or which were actively planning to do so. The Questions on which responses were invited are set out in Annex 3. The written evidence received to the end of October 2000 was published on 8 November as *Human Genetic Databases: written evidence*¹. The publication is also available on the House of Lords website (www.publications.parliament.uk/pa/ld/ldsctech.htm). This description of current and planned activities in human genetic databases, mainly in the words of the major players themselves, is a very helpful resource for the Working Group and others involved in this consultation.



¹ HL Paper 115, ISBN 0 10 411500 9, The Stationery Office, price £14.40, telephone orders 0845 7 023474

2. What is personal genetic information?

- 2.1 Personal genetic information may be defined as any information about the genetic makeup of a person. This information may be derived from a sequence of the components (the nucleotides, the As, Ts, Cs, and Gs of the genetic code) that make up the DNA molecules in the chromosomes which are found in almost every cell in our bodies. There is also DNA in the mitochondria (small independently reproducing organelles found in most cells which power the chemical machinery of the cells). The chromosomes, together with the mitochondria and other biochemical structures, provide our biological inheritance – our genome – which is transferred from generation to generation in the egg and sperm.



From "Your Genes, Your Choices", American Association for the Advancement of Science (1999)

- 2.2 The DNA of the chromosomes has some segments of sequences which we call genes – probably some 50,000 in all. Genes are involved in arranging the sequence of elements that make up all the proteins in our bodies – the building blocks and chemical messengers of our cells. While we all carry the same set of genes, there is some variation from individual to individual in the genetic code (gene sequence) within each gene. This variation may lead to variations in the structure of particular proteins which, in turn, are associated with an individual's characteristics (for example in eyes or hair colour, resistance or susceptibility to particular infections, or in the effectiveness, or otherwise, of a particular drug treatment). More rarely, an individual may have a more major variation in the sequence of a particular gene which means that the normal protein associated with that gene cannot be produced. In such a case a genetic disease is the likely result.
- 2.3 About 95% of the DNA in the chromosomes lies outside the genes. This DNA is much more variable between individuals than the genes themselves, so it provides a very effective way of identifying individuals or tracing family relationships. Unless we have a monozygotic ("identical") twin, the sequence of the DNA nucleotides will be unique to an individual. It is estimated that, on average, there may be a million DNA sequence differences between two unrelated people.
- 2.4 As we have described, personal genetic information can be obtained from a wide variety of sources. Some of these methods have been used for many years. For instance, family histories of genetic disease have long been used clinically for assessing a couple's chance of producing an affected child or by insurance companies to set premiums for those at risk of inherited diseases. In this paper we are primarily concerned with genetic information that is derived by analysis

of DNA or associated biochemicals (proteins and RNA). We use this distinction because it is these new sources of genetic information based on DNA technology that provide new possibilities and challenges. There is public concern about the extent to which existing safeguards and regulation of information may be effective in the face of what can be done with the new technologies.

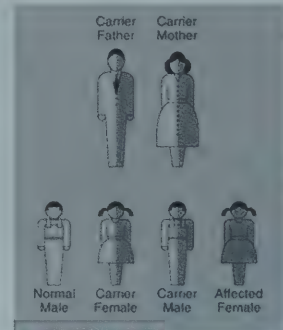
How can we obtain genetic information?

Information about a person's genetic makeup may be obtained in three ways:

- By taking a family history of a genetic disease
- By external observation of characteristics, such as eye colour
- By the analysis of blood or bodily tissue containing DNA, associated proteins or other biochemicals.

Family history

Genes pass through families. If a parent has a genetic condition, then there is a chance that this will be present in a descendant (or ascendant). Whether this is so will depend on the nature of the genetic condition. Family members may be affected in different ways: in the diagram one child has inherited the abnormal gene from both parents (and may be affected by the disease), while others are either carriers or are completely unaffected.



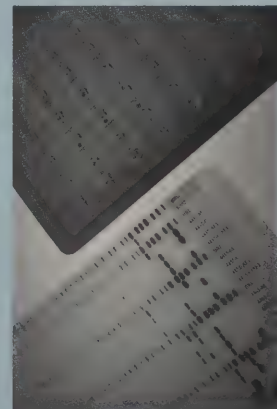
From "Understanding Gene Testing", National Institutes of Science, National Cancer Institute (1995)

External observable characteristics

It may be possible to draw conclusions about a person's genetic makeup by observing their appearance (eg certain eye colours are associated with particular variations in certain genes).

Analysis of blood or bodily tissue

When genetic information is obtained from the analysis of DNA (or proteins or other biochemicals closely associated with DNA) the sample may be obtained from any bodily tissue: a mouth swab, a few hairs or flakes of skin or a tiny spot of blood. It is the ability to base DNA or associated biochemical testing on these minute biological samples that makes the technology so powerful in crime detection. But by the same token, it could be very easy to obtain a sample for DNA testing from an individual without their knowledge or consent.



In almost all cases genes do not entirely determine the development of a disease and it is important to remember there is also interaction with environmental (eg smoking, diet and local) factors. Genetic information does not give us the full picture.

- 2.5 It can be argued that apart from the ease with which samples may be obtained for testing, and the prediction of some rare inherited diseases, there is little that distinguishes genetic information derived from DNA analysis from that which can be produced in other ways which have been available for many years. However, the ease of obtaining DNA and its analysis, in itself, raises fundamental issues. For example, a forensic database using DNA creates questions about the obtaining, storing and control of DNA samples (and the information derived from them) which are more complex than those posed by a collection of fingerprints. The situation is similar to the use of DNA techniques for determining paternity and other family relationships. For some inherited diseases new genetic testing techniques may provide much more accurate diagnostic and predictive information which can raise important questions about consent, confidentiality and the use of the information by family members and other third parties. As we learn more about the role of benign individual genetic variation in our responses to drug treatment and in disease susceptibilities, knowledge of an individual's genetic makeup will become important in a number of new contexts. Research exploring these important areas in itself poses questions about the use of genetic information from large groups of people.
- 2.6 It is perhaps also important to reflect on how un-special genetic information is. For example, a mother shares approximately 99.95% of her genetic information with her daughter but that same woman shares 99.90% of her genetic information with any randomly chosen person on the planet. There is only 1.5% difference at the level of genetic information between humans and chimpanzees, which means that while we share 99.90% of our genetic information with other humans, we share 98.5% of our genetic information with chimpanzees.
- 2.7 We also share 40-50% of our genetic information with bananas and cabbages. When thinking about our genes we need to remember that they are also shared with most of the rest of the 'living world'. Thus genetic privacy and the alleged rights to control "our" genetic information are to some extent illusory.
- 2.8 Over and above the issues raised by the new DNA technologies, it is increasingly recognised that DNA has a great symbolic significance for many people. It is widely described as the "blueprint for life" or the "essence of our humanity". While such phrases are not an accurate description of the biological role of DNA they have served to increase concerns about the use of DNA and the information derived from it.

Questions: What is personal genetic information?

- 2.1 **Is this a fair representation of what you believe personal genetic information to be? Is there anything else we should have added?**
- 2.2 **Do you have different concerns over the use of the 3 different sources of genetic information (family history, external observable characteristics, analysis of blood or bodily tissue) outlined above, and if so, why?**
- 2.3 **Should HGC be concerned with all sources of personal genetic information or should it mainly focus on the new DNA technologies?**

3. Is genetic information special?

- 3.1 Genetic information is a form of personal information, so why single it out for special treatment? If confidential biomedical information is already protected, then it could be argued that existing rules could simply be applied in the case of genetic information. This means that information about a person's genotype would be treated in exactly the same way as information about their blood pressure or childhood illnesses. If this were the case, then there would be no need to make any special recommendations as to storage, protection and use of genetic information.

Protection of personal information – the current situation

Society recognises the importance of personal information and protects it against improper disclosure or exploitation by others.

The principle of confidentiality is particularly emphasised in the medical context, where there are clear ethical and legal rules intended to discourage any breach of the patient's confidence:

- Codes of professional conduct. Such codes may embody respect for confidentiality, such as the General Medical Council's 'Duties of a Doctor: Confidentiality, 2000' for medical practitioners. The ethical requirement of confidentiality therefore may in some cases be enforced by professional discipline, which will strongly discourage the wrongful breach of confidentiality.
- Legal protection. The confidentiality of personal information is protected by the provisions of the *Data Protection Act 1998* and by common law remedies for breach of confidence. The *Data Protection Act* contains provisions for the secure handling of personal information which is either stored in computerised form or held in structured manual filing systems. Certain forms of genetic information (for example, which reveals racial or ethnic origins or which discloses information about physical or mental health) will come into the category of 'sensitive personal data' which are subject to stricter controls under the Act.

At common law, a person whose confidential information is wrongfully disclosed may bring an action for damages in respect of that disclosure. This may act as a deterrent to wrongful disclosure. There will also be situations where disclosure of personal information may be legal and desirable, for example for criminal

- 3.2 HGC has identified the following reasons that might suggest that genetic information warrants specific consideration:

- (1) Genetic information is uniquely identifying information. Only in the case of monozygotic twins ('identical' twins originating from the splitting of a single egg or zygote) will anyone have exactly the same genotype as oneself.

- (2) Genetic information can be obtained from a very small amount of material (the DNA in a single cell) and does not require lengthy observation or history-taking. It may also be obtained without the knowledge or consent of the person, either from a sample obtained in the past for another purpose, or from cells shed unknowingly (for example, from hairs).
- (3) Genetic information may be used to predict some rare inherited disorders that develop later in life and to predict what may be passed by a parent to children. This prediction may be made even before the birth of the person in question (cells may be taken from embryos and fetuses and preimplantation prediction is also feasible).
- (4) These predictive possibilities may be of interest not only to the subject of the prediction, but also to others, such as insurance companies or employers, who might wish to take them into account in their dealings with the subject. In many (though not all) cases, genetic information is not only information about the individual person, but about his or her biological relations. Genetic information about one family member may have significant implications for other members of the family, these include information about susceptibility to disease and issues of family genetic relationships. (In other cases, of course, genetic information may have no implications for other family members. This will be so where a mutation has occurred in an individual's cells leading to disease such as cancer.)
- (5) Genetic information has a potential commercial value and methods of obtaining it may be the subject of patents. In addition, there are complex issues as to ownership interests in human DNA.
- (6) Collections of genetic information combined with individual medical and life style information provides an important research resource for understanding individual susceptibilities to disease and its treatment. Increasingly drug treatments may be targeted to sub-sets of the population identified, in part, by genetic information.
- (7) Genetic information may be collected for a variety of very different purposes (disease prediction, determining relationship etc). However, once this information is collected, it is sometimes used for a quite different purpose from that for which the provider of the information originally gave consent.

Questions: Is genetic information special?

- 3.1 Is this a fair representation of the particular nature of genetic information? Is there anything else we should have added?**
- 3.2 Do you think that existing legislation and codes of professional conduct provide a sound basis for the protection of human genetic information?**
- 3.3 Does the protection of the confidentiality of genetic information require special consideration, or should it be treated in the same way as any other form of personal medical information?**

4. Ways in which personal genetic information may be obtained

- 4.1 Genetic information – in the sense identified above – may be obtained by analysis of an individual's DNA or associated proteins, or RNA. DNA analysis may involve the sequencing of the nucleotides in the DNA molecules; an examination of patterns of DNA fragments after the DNA is processed in the laboratory; or an examination of the proteins that the DNA produces. This may reveal individual variations within particular genes which are associated with genetic diseases or other individual variations. Identification of individuals by DNA is carried out by comparing it with the DNA from another sample collected from a known individual. A match will indicate that the two samples come from the same individual (or an identical twin). It is important to note that DNA in itself cannot be used to identify a person, this can only be done by comparison with a named sample. Identification of family (blood) relationships using DNA follows the same principle but involves analysis of degrees of similarity between the DNA samples being compared.
- 4.2 The genetic information thus obtained is distinct from the sample itself, and may be stored in a paper file or a computer. It may take the form of sequences related to the four letters representing the chemical bases that make up the DNA, or a picture (or numerical profile) of the pattern of DNA fragments. Thus, the full genetic identity of a person can be represented by a very large number of letters listed one after another in an extremely long sequence. A partial genetic profile would merely provide information about the sequence at particular points on a particular chromosome. It may show, for example, that a person has a particular mutation (a significant variation in the sequence of chemical bases). This mutation could be associated with a risk of developing a particular genetic disease.
- 4.3 Genetic analysis may be performed on a sample which the subject himself or herself has voluntarily provided for analysis purposes. Alternatively, it may be obtained from a sample that is obtained without consent (as may sometimes happen where a sample is taken by the police from a suspect). In other cases, it may be obtained from human cells shed or from the body (eg skin or hair), discarded by a person or covertly obtained (eg from a hairbrush). In addition, in paternity testing the DNA of individuals is compared to determine their biological relationship and this can involve complex issues of consent or indeed the issues of whether consent has been avoided.
- 4.4 Another important potential source of genetic information is collections of human tissue samples which have been made for other purposes, but which have been archived. The donor of these samples may not have given consent to analysis of the DNA they contain, and so this may be seen as non-consensual testing.
- 4.5 If the sample from which DNA is obtained is itself destroyed, the information derived from it still exists. This means that genetic information, which is easily preserved, could be communicated irrespective of what has happened to the original sample. It is also possible to draw conclusions as to the presence of a particular gene by testing for the presence within the body of a protein which is coded by that gene. In this way, genetic information can be obtained without specific DNA analysis.

5. Personal genetic information in medical practice: consent to genetic testing

Consent to tests (for treatment or advice)

- 5.1 It is an established principle in the law relating to medical treatment that a patient should not be treated without his or her consent if he or she is able to give it, except in special circumstances defined by legislation (for example mental health legislation). In general, incapacitated persons (such as those suffering from dementia or persons who are unconscious) may be treated without their consent if it is in their best interest to do so. This emphasis on consent reflects the importance given to the principle of autonomy, or self-determination in medical ethics. In general, a person who carries out a procedure without the consent of the patient may be liable in both civil and criminal law.
- 5.2 Consent must be informed. A person who consents to a procedure, the essential nature of which he or she does not understand, cannot be said to be giving a valid consent. The question of what information must be given to the patient in order for a consent to be considered informed has been the subject of a great deal of debate. There is a voluminous medico-legal literature on the subject and a considerable body of case law has grown up around the issue. In both English and Scots law, the test is that of what the reasonable doctor in the circumstances would deem necessary to disclose to the patient, although recent court decisions now confirm that there will be circumstances in which the court will depart from this standard in favour of the test of a standard of disclosure of risk determined by the court itself.
- 5.3 Consent must also be freely given. Attention should be paid to the possibility that a person has felt compelled to give a consent because of the constraints in his or her circumstances.
- 5.4 In the usual case, the patient will give either explicit or implicit consent to the carrying out of tests. A patient who permits a doctor to withdraw a sample of blood for the purposes of testing (the nature of the tests not being explained to the patient) will be assumed to be consenting. The basis of this is that patients in such a case understand that the procedure will involve certain tests which will be appropriate to the symptoms with which they are presenting. It may not be acceptable to assume consent without at least some indication of the scope of the tests. It may be, of course, that the doctor will give a further explanation of what tests are envisaged, but in many cases this will not be done for reasons of time and because the technicalities of the testing may be thought not to be information which the patient expects or requires.
- 5.5 Would the carrying out of a DNA analysis on a sample, for example a tumour biopsy, provided by a patient require the specific consent of the patient? If a doctor were to take a sample from a patient and say that it is to be subjected to "tests", would the patient's consent to this be considered valid, even if there was no disclosure of the fact that DNA analysis is intended?

Genetic testing in medical practice

In the medical context, genetic testing may be undertaken as part of the process of treating or advising an individual patient. There are four forms of genetic test in this category:

- (1) *Diagnostic genetic testing* – Use of genetic testing in individuals with symptoms to aid in his or her diagnosis, treatment and management.
- (2) *Presymptomatic genetic testing* – Primarily carried out in healthy people or those without symptoms to provide information about that individual's future health, with respect to specific genetic (also called inherited or Mendelian) diseases. Such a test result may indicate that the individual has a high likelihood of developing the disorder or of excluding it. Presymptomatic testing is most frequently used in late onset dominantly inherited diseases such as Huntington's Disease.
- (3) *Carrier Testing* – Used to detect individuals who possess a single copy of a gene which follows a recessive pattern of inheritance (eg cystic fibrosis, sickle conditions or thalassaemia). Individuals who are carriers will not develop the inherited disease but if they have children with another carrier, these children have a one in four chance of developing the disease.
- (4) *Susceptibility testing* – Gene variants have been discovered which are associated with common diseases such as Alzheimer's disease and diabetes. While the associations between carrying the gene variant and developing the disease do not appear to be close enough to make predictive testing useful, it is likely that increasingly genetic tests for such variants will be used to target drug treatments to those most likely to benefit from them. This rapidly growing field of genetically targeted drugs is known as pharmacogenetics.

- 5.6 There is no direct legal authority on this issue. Differing views have been expressed by commentators, with some arguing that the implications of genetic testing are such that specific consent should be required; the legal position being unclear, the safer view to take might be that the law requires specific consent. Such a view would be in harmony with the ethical consensus on the matter. Existing guidelines on genetic testing in the clinical context emphasise the need to inform the patient of the nature and implications of the test.
- 5.7 Genetic testing may also be undertaken in a research context in which the object of the test is to expand our understanding of processes involved in the development of diseases and their treatment. (Some forms of treatment, of course, may be conducted as part of a project of research.) We propose to deal with genetic testing in research separately, as it gives rise to particular issues.

Existing guidelines on consent to genetic testing in the clinical context

The Advisory Committee on Genetic Testing (ACGT) stated in its *Report on Genetic Testing for Late-Onset Disorders* (1998): “When an individual is able to give consent, specific freely given consent is required before testing. In the case of presymptomatic genetic testing of healthy individuals, written consent should always be obtained, though written consent is not in itself a substitute for careful face to face explanation.”

Article 12 of the Council of Europe’s *Convention on Human Rights and Biomedicine* (1997), which deals with predictive genetic tests states that these tests, which can only be performed under the article for health purposes or in the context of health research, must be “subject to appropriate genetic counselling”. Furthermore, the Explanatory report to the Convention makes it clear that “proper informed consent” is “of particular importance” (para. 80).

The Committee of Ministers of the Council of Europe in 1992 adopted a Recommendation on Genetic Testing and Screening for Health Care Purposes (No. R(92)3). This states, in principle 5, that genetic testing, even when offered systematically, “should be subject to ... express, free and informed consent”. Further, the principle emphasises the importance of respect for individual self-determination. These general principles suggest that a specific explanation of the fact that a test involves genetic analysis is called for.

UNESCO’s *Universal Declaration on the Human Genome and Human Rights* (1997) also emphasises in Article 5b the importance of prior and informed consent of the person concerned to any diagnostic test concerning an individual’s genome.

The ACGT considered genetic paternity testing in 1998 and made recommendations about quality assurance and accreditation systems as well as the importance for consent from the mother or third party with parental responsibility. It recommended that these principles should be incorporated into a code of practice. A voluntary Code of Practice on Genetic Paternity Testing is currently under preparation.

- 5.8 A particular consent issue arises in relation to future developments in high throughput testing devices that provide results for a large number of DNA sequences (gene chips). The development of such tests might make it difficult to focus on specific chromosomal sites and exclude other information. This might require the agreement of protocols relating to the use of such devices.
- 5.9 In consenting to a genetic test, a person may be assumed to consent to being told the result, although a person may specifically ask not to be told (intending the result to be for the clinician’s use). This consent to the receiving of information, however, does not cover information unconnected with the specific test to which consent has been given. If genetic analysis reveals additional information about some other condition, the question arises as to whether this information may be communicated to the patient. There would appear to be a strong case for communicating this to the patient if treatment were available or if the patient could take some other action (such as changing diet) to reduce the

- risk of developing or aggravating a condition. This suggests that the possibility of additional information being revealed should be raised with the patient before the test is undertaken and the patient's views sought, rather than having to decide after the event whether the patient would or would not want to know.
- 5.10 Issues arise as to the placing of the results of a genetic test on the patient's record. Normal practice is for a GP to add the results of tests to a patient's notes. A patient could, however, ask for the results of a test carried out by hospital testing services not to be sent to his or her GP. Some testing services – such as those carried out for sexually transmissible diseases – may be offered anonymously, but this is not the practice in clinical genetic services. The entering of information on a record against the patient's wishes raises legal and ethical issues.
 - 5.11 The interests of children and those adults who are incapable of giving consent will need to be considered. The carrying out of a genetic test as an aid to the diagnosis of a symptomatic condition may be considered part of the normal treatment process, and therefore a matter to which, in the case of children, parental consent may quite appropriately be given. However, presymptomatic genetic testing of children for late-onset conditions raises more complex moral questions. This issue was considered by the Clinical Genetics Society Working Party on the Genetic Testing of Children (1994) and the Advisory Committee on Genetic Testing (ACGT) in its *Report on Genetic Testing for Late-Onset Disorders* (1998). The Committee recommended that the testing of young children for such conditions for which no presymptomatic treatment can be offered should not be undertaken. In the case of adolescents who have the capacity to consent, the Committee suggested that testing might be undertaken after full explanation and discussion of the implications.
 - 5.12 There may be cases in which a genetic test is deemed appropriate in a clinical context but parents are unwilling to give consent. These situations are essentially no different from other cases in which there is an issue of parental consent to treatment, and should be resolved by the same legal rules. English and Scottish law both recognise the ability of children over the age of sixteen to give an independent consent to medical treatment. In the case of children under the age of sixteen, a valid consent to necessary medical treatment may be given by the child provided that he or she is capable of understanding the issues involved (the Gillick test). The question of a young person's ability to understand the issues involved in genetic testing would depend on the psychological implications of the test, and this would suggest that extreme caution be exercised in reaching any conclusion as to competence.
 - 5.13 Children are entitled to have their medical information treated as confidential. However, it will obviously be necessary for parents to be informed of medical facts (including genetic information) about their children as long as this information is needed by them in order to carry out their parental duties in relation to their child. In the case of a child nearing maturity, this may involve a delicate balancing of a young person's desire for privacy against the legitimate interests of parents in the welfare of their child.

- 5.14 The involvement of children in genetic research, which has no therapeutic implications for them (non-therapeutic research), raises difficult legal issues which have yet to be resolved. The courts in both England and Scotland have not pronounced on this issue, and there has been a lively and prolonged debate on the ethical implications of this. Such involvement is probably legal provided that there is a negligible risk for the child and there is no reason for the research to be considered against the best interests of the child. Parental consent should be obtained.

Paternity testing

- 5.15 There are some cases, such as paternity testing or forensic testing where special consent arrangements may apply. Forensic testing is covered in Section 10. In paternity testing, additional issues of consenting on behalf of children apply.
- 5.16 DNA testing provides an accurate means of determining parentage. A sample obtained from apparent parents may be compared with that of the child and a conclusion reached as to biological parentage. In the UK, a large proportion of paternity testing is carried out on behalf of the Child Support Agency (CSA) to determine child maintenance. It is also frequently used in immigration cases.
- 5.17 This procedure requires the consent of any adults involved in it; an adult may refuse consent, but this may justify a court (or, from next year, the CSA) making an adverse inference from this refusal. In the case of the testing of a child under the age of sixteen, consent must normally be obtained from the person with parental responsibility for that child, but the consent of the child himself or herself is not required. Courts have the power to direct the carrying out of tests, and in exercising this power the court will take into account the welfare of the child (although other considerations may justify the directing of tests). In general, courts have placed great weight on the consideration that the truth as to parentage should be known. Under recent legislation (the *Child Support, Pensions and Social Security Act 2000*) the court has power to waive the consent of a person with parental responsibility for a child if it believes that the test is in the child's best interest. The position in Scotland under the *Law Reform (Miscellaneous Provisions) (Scotland) Act 1990* is broadly similar to the position in England on court requests for the taking of DNA tests.
- 5.18 A major issue of principle in relation to the use of DNA testing in disputed paternity cases is that of the right to refuse a bodily sample for DNA profiling. The law recognises this right, even if its exercise may lead to an adverse inference being taken by the court. The civil liberties argument against coercion in this area is therefore acknowledged, although children under sixteen may be tested without their consent. Advances in the technology of testing means that paternity testing is becoming easier. Until recently it was necessary to take blood samples from both parents to establish parentage. It is now possible to produce accurate results from cells taken from inside the mouth using buccal swabs, hair follicle samples or from finger prick blood samples. This enables paternity testing to take place "over the counter" using sampling kits advertised by non-UK organisations via the Internet and sent through the post.

- 5.19 Additionally, although routine paternity testing is carried out with samples taken from the mother, putative father and the child, increasingly tests are being undertaken using samples provided by the child and the putative father alone (“motherless testing”). The ACGT recommended in 1998 that the mother, or a third party with care and control of the child, should give written consent to testing before a “motherless” test is performed. Sometimes these tests may be performed overseas, which raises issues as to whether the complex legal issues involved in whether the putative father has the right to consent to the testing of the child can be properly addressed.

Mentally incapacitated adults

- 5.20 In the past, the feelings and indeed the rights of persons with a mental incapacity were not given adequate protection by society in general and by the law. There is now a much greater sensitivity to the interests of this group, and there has been considerable ethical and legal debate as to how to enhance the level of protection afforded. In Scotland, there has recently been a thorough revision of the law in this area. The *Adults with Incapacity (Scotland) Act* 2000 has introduced a more accessible and responsive system for the supervision of the affairs of those who cannot perform this task themselves. This legislation also provides a system for the protection of the interests of incapable patients in both a treatment and research context. The details of this system are beyond the scope of this document, but the underlying principle is that reasonable treatment may be given under the authority of the Act and that, where a guardian or welfare attorney has been appointed, there should be consultation with the guardian or attorney. Participation in research may be allowed, provided that stringent conditions are met. It would be possible under this legislation for a research-oriented genetic test to be performed on a person who is incapable of consenting, provided, among other conditions, that the consent of a guardian, welfare attorney, or nearest relative has been obtained and the research cannot be carried out on a person who is capable of consenting.
- 5.21 In England and Wales, when an adult lacks capacity to give consent, no-one else is entitled to give consent on behalf of the individual. In general it will be lawful to carry out tests on an individual if such tests are in the best interests of the person concerned, for example to assist in establishing the cause of symptoms. It might also be in the best interests of an individual to have a presymptomatic genetic test if as a result treatment could be given, or environmental modifications made, such as altering the diet, to make it less likely that the disease would develop in the future. Whether or not it is in the best interests of an individual to be tested presymptomatically for an untreatable disorder would require very careful consideration of all the relevant circumstances. In all cases, if it is thought that the person’s incapacity will be temporary, any genetic testing should be delayed until consent is possible unless it is essential in the individual’s care and treatment.

Family linkage studies

- 5.22 When the molecular basis of a disorder has not been established exactly, it may be helpful to conduct “linkage studies” involving the testing of several members of the same family. Linkage analysis is a statistical method for detecting linkage between a disease and markers of known location by following their inheritance in families. If such linkage can be established within a family, it will be very likely that a family member who is found to have inherited the known marker will also be likely to have inherited the disease that appears to run in that family.
- 5.23 An adult who is capable of giving consent is able to consent to giving a blood sample for linkage analysis, even though the test may be of no benefit to themselves, after having received appropriate information. But although it is in the best interests of the family as a whole for as many people as possible to be tested when trying to establish linkage, the involvement of children and adults incapable of giving consent raises important concerns.
- 5.24 In the case of children, the English courts have recognised that a person with parental responsibility can consent to an intervention which, although not in the best interests of the child, is not against the interests of such a child. On that basis, it is possible that it would be lawful for a child to participate in linkage analysis with consent from a person with parental responsibility, although such a decision has to be made individually on the basis of the particular situation. The courts have not considered a case on this issue in relation to adults. Such a test would be lawful if it was in the best interests (which is not confined to the best medical interests) of the adult concerned. In their 1995 report on Mental Incapacity, the Law Commission briefly considered this issue. Although they concluded that it was not necessary to clarify the law on this issue at the time of their report, they did conclude that it might be appropriate in the future for the law to be reformed (subject to the will of Parliament) to make lawful procedures – such as linkage studies – which although not carried out for the benefit of that person, will not cause him or her significant harm and will be of significant benefit to others.

Prenatal genetic testing

- 5.25 Prenatal genetic testing was considered by the ACGT in its report for consultation, *Prenatal Genetic Testing* (2000). The availability of prenatal genetic testing entails issues of the proper use of genetic information: such information may play a part in a decision as to whether or not to continue with a pregnancy. The Committee made a number of recommendations, including ones relating to the obtaining of proper consent. Pre-implantation genetic diagnosis, a procedure that involves IVF treatments, similarly raises profound issues of the use of genetic information to determine which embryos will be given the chance of further development. This issue was discussed in a joint ACGT/Human Fertilisation and Embryology Authority consultation document published in November 1999.

Questions: Consent to genetic testing

- 5.1 Is this a fair representation of the consent issues in genetic testing?
Is there anything else we should have added?**
- 5.2 How much information do you think is required for the informed consent of an adult in the following cases:**
 - (1) diagnostic testing;**
 - (2) carrier testing;**
 - (3) presymptomatic genetic testing;**
 - (4) testing carried out in pregnancy?**
- 5.3 Is it acceptable for family linkage studies to be carried out on:**
 - (1) children with consent from a person with parental responsibility and if so, under what conditions?**
 - (2) adults not capable of giving consent for the benefit of other family members and if so, under what conditions?**
- 5.4 If testing techniques give information on many genes or diseases, then should all the results be communicated to the patient, or only those to which the patient had explicitly consented?**
- 5.5 Would the carrying out of a DNA analysis on a sample, for example a tumour biopsy, require the specific consent of the patient or would general consent for medical tests to be conducted be sufficient?**
- 5.6 How can the principle of informed consent be applied for paternity testing or other testing conducted by organisations based outside the UK?**

6. Protecting the confidentiality of personal genetic information: communication within families and the right to privacy

Communication within families

- 6.1 Genetic (inheritable) diseases are, by their very nature, not individual matters and may have consequences for family members. If a genetic test is performed on an individual or a genetic risk assessment is made, the information so obtained may have implications for other members of the family (but not in the case of common genetic variations or diseases, where the information may not be usefully predictive). Often family members will be aware of the possibility of an inherited condition running in the family. When a member (or members) of the family has been to a consultation, they will in many cases pass on the information they have received to other relevant family members, who in turn may decide to seek clinical advice for themselves. Clinics may support and indeed encourage the process of family communication and genetic counsellors may give advice on how such information may be disseminated to those for whom it may be most relevant. It is not uncommon, though, for people to find it difficult to talk to others in the family about the problem, particularly if a family has been unaware that a condition is inherited.
- 6.2 It is a basic approach of clinical genetics that, in most situations, those who carry significant risk of inherited disease for themselves or for any children they might have are better off knowing about such risks. Occasionally there is a difference of view between genetic counsellors and the person consulting the counsellor as to whether or not other family members should be told about a condition. If there are actions other family members could take which might reduce the risks to themselves or their children, counsellors usually point this out. Best practice is to give counsellees letters describing the situation which can be used as the basis for passing information to other family members. In some cases, this advice will be rejected, and the question then arises as to whether counsellors themselves should pass on the information. This would amount to a breach of the principle of confidentiality – but are such breaches justified in the circumstances?
- 6.3 In one view, the confidentiality of medical information should be given absolute protection and should never be breached, even if the passing on of the information would enable others to protect themselves from the risk of harm. This view enjoys some support, particularly among those who feel that medical confidentiality has been weakened in recent years. A more widely-supported view – one which is endorsed by the General Medical Council, for example – is that it may be permissible to breach medical confidentiality in order to protect another from serious harm. Such breaches would only be made if the person in possession of the information had tried unsuccessfully to persuade the patient to agree to its disclosure and if, on balance, the harm caused by non-disclosure is thought to outweigh the harm caused to the patient by breaching confidentiality. In any event, the information disclosed should always be limited to that which is strictly necessary to prevent serious harm (Health Services Guidance HSG(96)18: *The Protection and Use of Patient Information*).

- 6.4 A particular moral issue which must be taken into account in genetic cases is the fact that information passed on to other family members by a counsellor – or indeed by a family member himself or herself – may not be wanted by the person to whom it is disclosed. If recognition is given to a “right not to know”, then this right would be infringed if a family member were to be given information that he or she had not requested. It is for this reason that some ethics committees have taken the view that in the research context no unsolicited approaches should be made to those at risk.
- 6.5 Even if there is disclosure, it should not be assumed that this would result in any action on the part of the person receiving the information. The fact that there may be no response, or that the information may not be wanted, makes it less easy to say that the disclosure is always going to cause less harm than non-disclosure.
- 6.6 Legally, a breach of confidentiality may give rise to an action for damages on the part of the person entitled to confidentiality. British courts have recognised, however, the legitimacy of breaching confidence in order to protect others. This involves a balancing of interests to determine whether a public interest in disclosure outweighs the private interest in keeping the information confidential. The law on confidentiality is common (that is, non-statutory) law. There is currently no legal precedent in the United Kingdom touching upon the breach of confidentiality in relation to genetic information, and it could be argued that this leaves the genetic counsellor in an uncertain position. However, unless the legislature or the courts were to endorse the principle of absolute confidentiality – which would at least lead to certainty – the circumstances in which a breach would be justifiable would have to be stated in general terms, as every situation would be different and would require moral evaluation which took account of the particular features of the case.
- 6.7 There has been some legal recognition in the United States of a duty on the part of genetic counsellors to disclose information to those who are potentially at risk of harm. Such an approach is based on the notion that a genetic counsellor owes a duty of care to the family members of the patient or client. There are no precedents directly on this point in the United Kingdom, but, on the basis of current understandings of the law of negligence and analogous cases, it is unlikely that the British courts would infer a duty of care in such cases. This is a complex area of law, however, which the courts are still developing, and it is possible that the boundaries of liability could be extended to impose liability for failure to warn of genetic risk.

The right not to know

Does the individual have a right not to know genetic information about himself or herself? Such a right has attracted strong support in the debate on the ethics of genetics, and indeed it has been recognised in international declarations and conventions. The United Nations' *Universal Declaration on the Human Genome and Human Rights* (1997), for example, provides in Article 5 for the protection of the right to decide whether or not to be informed of the results of any genetic test. Similarly the Council of Europe's *Convention on Human Rights and Biomedicine* (1997) states in Article 10(2) that the wishes of those who do not wish to be informed of information collected about their health should be observed.

The argument in favour of a right not to know is based on the autonomy of the individual. The desire not to know whether one is at a genetic risk of developing a particular condition may be a strong one, and it may be that the individual concerned feels that genetic knowledge would bring with it an unacceptable degree of anxiety about the future, particularly where there is no treatment available for a particular condition. If a person has decided that it will be better not to know, for example, that he or she has a heightened chance of developing a life-threatening disease, then that decision must be respected, even if the person in question could do something to avert the onset of the disease. This is because, in general, we reject paternalism in the medical context.

The right not to know has not been specifically recognised by the courts in the United Kingdom, but it might be possible to establish it on the basis of Article 8 of the European Convention of Human Rights, which recognises the individual's right to private life. This right, however, is subject to possible restriction in the interests of health and social policy. Clearly this involves a balancing of interests, but it would be difficult to imagine circumstances in which the wider public interest would justify forcing genetic information on an individual who does not wish to receive it.

As the predictive power of genetic testing increases, it is likely that concern will grow over the extent to which genetic knowledge weakens the element of "openness" which we see in our future. The ethical issue that must then be addressed is this: to what extent should we – and can we – protect people from unwanted knowledge about their genetic status and their personal future?

The right not to know does not find universal support. Critics of this concept argue that what is being asserted is a right to ignorance, a concept which may be more difficult to endorse. In particular, the argument is made that people cannot be expected to make autonomous choices about their future without having all the relevant information at their disposal and that declining to give them this information is indefensibly paternalistic. In this view, there is a presumption that knowledge is to be preferred to ignorance, particularly when matters of reproductive choice are involved.

Confidentiality, Privacy, and Human Rights

- 6.8 It is clear that people consider genetic information about themselves to be significant information that requires legal protection. Although we clearly have some kind of interest in our DNA, how exactly can this be defined? One way of describing this interest is to use the term genetic privacy. This implies that our interest in our genome is a value of the same sort as our interest in being protected against personal intrusion of other sorts.
- 6.9 The law on breach of confidence provides some protection for this interest. The law provides a remedy in those cases where a person is entrusted with confidential information and then divulges it to another who is not entitled to receive it. However, this remedy is not available where there is no legally relevant relationship between the parties. If the confidential nature of genetic information were to be given broader protection, then this would probably have to come through the wider development of a law of privacy, either through judicial decisions or through legislation.
- 6.10 The language and institutions of human rights might provide a useful way of enhancing protection for this sense of the private nature of genetic information. Human rights law has evolved extensively over the last few decades and has now entered a new phase of development in the United Kingdom with the coming into force of the *Human Rights Act* 1998 (which introduces into domestic law in the United Kingdom those rights recognised in the European Convention of Human Rights².) It is not yet apparent how the British courts will apply the general principles enunciated in the Convention, but the general pattern of development of human rights law in Europe since the promulgation of the Convention is clear enough. Under Article 8 of the Convention (which protects the right to private life) it could be possible to assert a right to the confidentiality of genetic information, and if this were to be recognised, then acts of the authorities which compromised this could be deemed to be in contravention of Convention rights.
- 6.11 The precise contours of this right would need to be identified. A possible testing ground would be matters such as the right of the State to retain indefinitely DNA information in forensic databases. Here the balance would be between the right of privacy recognised in Article 8 of the Convention as against the permissible restrictions of that right embodied elsewhere in the same article. These restrictions provide that privacy may be limited in pursuit of certain purposes, one of which is the administration of criminal justice. (Through the data protection principles, the *Data Protection Act* 1998 puts restrictions on the length of time for which personal data may be kept. The rule is that they must not be kept for longer than is necessary for the purpose for which they are processed. This restriction does not apply, though, where the data are processed only for research purposes provided that the conditions set out in Section 33(1) of the 1998 Act are met. There are also more limited derogations on a case by case basis, including for the prevention or detection of crime or the apprehension or prosecution of offenders.)

2 The Council of Europe's *Convention for the Protection of Human Rights and Fundamental Freedoms*' (1950, subsequently amended)

- 6.12 Another area in which the right of privacy might be considered is in genetic screening programmes. Genetic screening programmes involve approaching persons who, through the taking of a family history, have been identified as being at risk. Approaching such persons involves drawing their attention to genetic information which they have not requested and is, in a sense, an intrusion into their privacy. The clear benefits of screening programmes for serious illnesses (such as cancer) mean that it is unusual for people involved to object to such approaches; however, if genetic screening were to become more common and to be offered for less serious considerations, then it might be possible to argue that the intrusion upon privacy is unwarranted and less easily defended. Again the matter becomes one of balancing the right to a private life against reasonable expectations that an individual will welcome the information and the opportunity which it may bring to avoid or mitigate a risk to health.

Questions: Confidentiality of genetic information within families

- 6.1 Is this a fair representation of the issues surrounding confidentiality with relation to families? Is there anything else we should have added?**
- 6.2 In what, if any, circumstances, should the non-consensual disclosure of genetic information be allowed within a family setting?**
- 6.3 Does the current framework of law against the unauthorised disclosure of medical information provide adequate protection for genetic information?**
- 6.4 What further measures, if any, should be considered to give particular protection to the confidentiality of genetic information in this context?**
- 6.5 In the family context, should there be a “right not to know”? If so, should this right be absolute, or could it be breached in certain circumstances? If it could, what should the circumstances be?**

7. Personal genetic information in research

- 7.1 Medical progress has throughout history relied on learning from human beings, their bodies and human tissue samples. Over recent years human organs, tissue and genetic material has been collected and stored and this vast resource throughout the world, which could be described as “the human tissue archive” has enabled countless crucial medical discoveries to be made. It is clear that if we, humankind, continue to add to this archive and it remains available as a research resource the benefits to humankind could be very significant.
- 7.2 Of course, contributions to this “archive” must be made with scrupulous ethical and legal safeguards, and this document is largely concerned with these safeguards and the respect for human individuals and human rights that these safeguards represent. However, it is important to be clear that unless human tissue and human genetic information continues to be gathered and made available for research there will be costs in terms of prolonged human suffering from disease.
- 7.3 It is important for the public to understand these potential benefits and why doctors and medical scientists wish to obtain samples in the first place. It is also important that appropriate consent is obtained from research participants beforehand. Some examples of important research discoveries are given below.

Examples of medical discoveries from the “human tissue archive”

- The discovery of the link between smoking and lung cancer was made through painstaking examination of pathological samples obtained from the bodies of autopsied smokers.
- A leading cause of death in many countries is atherosclerotic cardiovascular disease, the most common form of heart disease. The connection between heart disease and blood pressure, cholesterol and high-density lipoprotein and diet was due, in large part, to research on the bodies of Korean war casualties.
- The availability of banked DNA specimens from patients with familial adenomatous polyposis as well as frozen specimens of colorectal cancers, led to the identification of the APC (adenomatous polyposis coli) gene. Mutation of this gene or other connected genes is implicated in most colorectal cancers.
- For many cancers, important new information about the genetics of these diseases depends on retrospective studies of human tissue.

Examples taken from David Korn M.D. “Research Involving Human Biological Material: Ethical Issues and Policy Guidance Vol II: Commissioned Papers”, National Bioethics Advisory Committee (2000).

- 7.4 To make best use of information from the human genome project, researchers need to understand how variation in our genes (at the level of the DNA) influences our biology: how people respond to environmental differences, to infections, to nutritional differences. The aim is to be able to understand in more detail why some people get asthma or hypertension, others succumb to colds more frequently and yet others get fat or stay thin on very similar diets. If researchers can begin to understand the biology, then they might be able to devise ways to prevent or more effectively treat these common problems.
- 7.5 Only if researchers can collect genetic and clinical information on a very large scale, are they likely to get answers to some of these questions. For this, researchers need access to many DNA samples and some ethical way to link them to longitudinal (ongoing) clinical information. Researchers do not need access to the identity of the patient, only to know that the right DNA is linked with the correct records. It will therefore be important to use (and further develop) techniques to protect individual privacy.
- 7.6 The research will mean looking for answers through large-scale data analysis by computers without knowing what the actual questions are. Researchers are looking for links and correlations that cannot necessarily be foreseen at this stage. From those links, they hope to learn more about how the diseases develop and how they may be prevented. Thus when considering the giving or withholding of consent to the donation of tissue or organs for research it is important that research participants bear in mind that uses beyond those immediately apparent might enable these samples to contribute to important advances in both knowledge and therapy.

Volunteers and consent

- 7.7 The importance of obtaining the consent of research participants who are able to give it is stressed at both international and national level and indeed could be described as one of the fundamental principles of ethical research. At the international level, relevant documents include the Council of Europe's *Convention on Human Rights and Biomedicine* (1997), Article 16 of which specifies a need for express and specific consent to participation in research. At the national level, ethics committees enforce the ethical requirement of consent, and all statements of professional bodies (such as Royal Colleges) and funding bodies (such as the Medical Research Council; MRC) stipulate the need for the informed consent of the research subject, if the subject is capable of giving such consent. The issues have been covered in depth by the MRC in its draft guidelines, *Human Tissue and Biological Samples for Use in Research* (1999). The issues of consent and the information to be given to participants in genetic research are addressed in the ACGT's *Advice to Research Ethics Committees* (1998).
- 7.8 Genetic research is subject to the same standards as any other branch of medical research in that informed consent is required from the donor at the time a sample is obtained. There are, however, certain features of genetic research which give rise to special ethical issues. One of these – the possibility of doing further research beyond that for which consent was obtained – is also faced in any form of research which involves work on stored samples rather than the carrying out of procedures on a volunteer. This is not unique to genetics, of course; nor is it an entirely new problem. There has been much discussion on the moral issue surrounding the conduct of research on samples originally taken for pathology

testing, many of which are stored in archives and could be useful for research in ways that could not have been envisaged at the time the sample was taken. DNA might also be obtained from a donated sample or from leftover surgical waste.

- 7.9 Where a sample is given purely for research purposes, the question arises as to whether the donor of the sample has any continuing interest in that sample. One view might be that the donor makes a gift of the sample (either unconditional or with conditions specifying what it might be used for). An unconditional gift entitles the recipient to do what he or she wishes to do with it, in the same way as the recipient of an ordinary gift may use the gift as he wishes. In this view, DNA gifted to a researcher could be subjected to whatever processes the researcher chose to employ. It is implied from this view that the researcher could, in the absence of any agreement to the contrary, use information derived from the analysis for a variety of purposes. However, the Data Protection Act states that personal data may only be used for the purposes for which they were collected or for other compatible purposes.
- 7.10 This view conflicts with current moral opinion, the clear direction of which is to recognise that the donor has a moral interest in what is done with bodily samples. (See, for example, the guidelines of the Royal College of Pathologists in *Guidelines for the Retention of Tissues and Organs at Post-mortem Examination* (March 2000).)
- 7.11 What are the limits of this moral interest? Does it extend no further than agreement that the sample may be used for research (in general), or is it necessary to agree to a particular form of research? Again we are faced with the issue, discussed above, of the “special nature” of genetics and whether specific consent is required if any genetic analysis is to be conducted. The reasons for requiring specific consent in the context of treatment are clear enough (see Section 5), but do the same considerations apply even if the sample is to be anonymised (see box below) and there is no possibility of feedback to the donor? The generally accepted answer to that is probably not, but this does not mean that the donor of the sample may not have an interest in what happens to the sample. Certain information may be revealed by DNA analysis, and even if this information will be anonymised and therefore not in any way be associated with an identified individual, it is possible that some people may feel protective of that information. They may, for example, be concerned about the effectiveness of anonymisation. Requiring specific consent to genetic research would allow such persons to decline to participate if they felt these concerns.
- 7.12 Another aspect of this moral interest is the stake that the donor has in knowing what sort of genetic research is contemplated. In some cases the potential usefulness of the sample for a particular type of research cannot be envisaged when the sample is taken. It may be that the donor will object to certain forms of research for religious or other reasons. Should such possible objections be accommodated? A possible view is that as long as the research is otherwise ethical, no account should be taken of any further, individual objection. This is open to the objection that a failure to alert a potential donor of a factor which might be expected to influence his or her decision may render a consent invalid.

Definitions of types of tissue sample

Anonymous (unidentified) samples come from unknown or unidentifiable sources. There is no way of tracing them to any particular person.

Anonymised (unlinked) samples previously had individual identifiers (proper names or codes) but these have been deliberately removed before giving the samples to the researchers.

Coded ('linked' or 'identifiable') samples can be matched with individuals through a numbered code. Access to the code linking numbers to names is restricted to a small number of people, researchers receive only the number, not the names.

Identified samples contain either a name or a patient number that researchers can match with a name.

An alternative terminology is used in the MRC's *Guidelines on Personal Information in Medical Research* (October 2000):

Unlinked anonymised data contains no information that could reasonably be used, by anyone, to identify people.

Linked anonymised data is anonymous to the people that receive and hold it (eg a research team) but contains information or codes who would then allow others (eg those responsible for the individual's care) to identify people from it.

Coded data is identifiable personal information in which the details that could identify people are concealed in a code, but which can be readily de-coded by those using it. It is not anonymised data.

- 7.13 In its advice to research ethics committees, the ACGT considered that consent must have been obtained for each genetic test, unless the new test is covered within the scope of the original consent. The exception to this advice is where the study is conducted in an anonymised fashion. ACGT also considered that research involving multiple gene tests is no different from research involving a single gene test. It is important to note that ACGT's advice primarily relates to genetic tests that have known or predictive clinical value. The majority of genetic research results will not have, and may never have, clinical or predictive value at the time they are obtained.

Large-scale genetic databases linked to health records

- 7.14 It could be argued that there is a need to require consent to genetic research as such, without further specification. It could be seen as unduly restrictive of research to require that consent be given to the performance of named tests, bearing in mind that a large sample of volunteers will be needed for meaningful studies (such as that contemplated in the MRC/Wellcome proposal to establish a large genetic database in the United Kingdom) and work will focus on a range of conditions. On the other hand, considerations must be given to the possibility that some people may have serious objections to the examination of their DNA for mutations suspected of being linked with a condition which they feel may be associated with stigma or potential discrimination. There is evidence to suggest, for example, that more people object to research into genetics and behaviour than object to research related to physical disease. One solution to this issue, which is discussed in the draft MRC guidelines on Human Tissue, is that the donor of a sample could be asked to consent to a particular type of genetic research (such as that concerned with heart disease or cancer). This would mean that there would not have to be a detailed specification of the particular tests involved while at the same time account could be taken of possible objections to certain types of research. An alternative system might offer the possibility of opting out of certain forms of research to which the donor had a particular objection. However, it should be borne in mind that it is difficult to “compartmentalise” medical research: research in one field will often have a close connection with, and impact on, research in other fields.
- 7.15 A further issue arises in connection with commercially sponsored research. Some people are prepared to participate in government or academically-funded medical research, but may have misgivings about participating in research which may lead to commercial gain. The MRC has considered this in its draft guidelines on Human Tissue. It takes the view that it is essential that participants are told that their sample may be used in research which could eventually lead to the development of commercial products and that they would usually have no share in the benefits of such use. However, if the research leads to a treatment for their disease, then the participants may benefit from the availability of that treatment.
- 7.16 There is a case for requiring donors to be informed of certain technical details surrounding the research. Not only is this likely to be important in ensuring maximum openness about what is being done, but it could be argued that it is essential to ensure that the consent is properly informed. The participant should therefore be informed whether the samples are to be anonymised, coded or linked, or identified. One reason for providing this information is that a participant might argue that she or he would not have consented had she or he known, for example, that the samples were not going to be irretrievably anonymised.
- 7.17 A separate Inquiry into human genetic databases is being conducted by the House of Lords Select Committee on Science & Technology. Details are in Annex 3.

Historical collections of material

- 7.18 All of the above applies primarily to future collections of material. The issue of historical collections also needs to be addressed. The MRC suggests that there will be circumstances in which it will be legitimate to use such collections if consent cannot be obtained. In the MRC's view, this will in general be so where the samples have been coded or anonymised and there is no potential harm for the donors of the material, either individually or as a group. In relation to genetic research, the draft Human Tissue guidelines state: "where a genetic test is of known predictive value, or gives reliable information about a known heritable condition, samples must be irreversibly anonymised before testing unless specific consent is obtained". Where the predictive value of a test is not known, non-consensual research is permissible on securely coded samples "provided that there is a strong scientific justification for not irreversibly anonymising the samples".

Feedback to participants

- 7.19 This is a controversial area. Where samples are anonymised, the link between the donor and the sample is broken and there will be no possibility of conveying to the donor any information that has been generated. Where, however, the sample is coded it will be possible to communicate the results of any test to the donor. The MRC recommends in its draft Human Tissue guidelines that this issue should be decided before the research starts. Donors should be asked what sort of information they would wish to have communicated to them; it may be that they will not wish to hear some sorts of information but be receptive to other sorts. They may not wish to hear, for example, about late-onset conditions for which no treatment may also be provided (susceptibility to earlier onset of Alzheimer's Disease may be an example of information that may not be wanted). The existence of heightened genetic predisposition to a condition that might be treatable or avoidable would be information of a very different nature.
- 7.20 There are other issues in relation to the difference in quality standards in research versus clinical practice, as well as differences in arrangements for consent, counselling and information provisions. Some of these have been addressed in the ACGT's *Advice to Research Ethics Committees* (October 1998). In principle, those receiving feedback from research should be treated in the same way as they would be in the clinic. For predictive information they should be offered genetic counselling to decide whether or not they wish to receive feedback and, if they do, the procedures should follow the usual clinical protocols.
- 7.21 The Council of Europe Recommendation (No. R(97)5) on the protection of medical data covers feedback of unexpected findings of genetic analysis if certain conditions are met. These relate to domestic law, consent and the likelihood of the information causing serious harm to the health of the individual and that of his or her family. Unless domestic law prohibits the giving of such information, the person should also be informed of unexpected findings of genetic analysis if this information is of direct importance to him or her for treatment or prevention, even if the person him or herself has not asked for this information.

Questions: Personal genetic information in research

- 7.1 Is this a fair representation of the issues in research? Is there anything else we should have added?
- 7.2 What types of information need to be given to someone donating tissue for use in a genetic research project?
- 7.3 For future research on the same sample, is specific consent to particular types of genetic research (eg research on heart disease or cancer) adequate?
- 7.4 Alternatively, would an “opt-out” system be acceptable and if so on what basis?
- 7.5 Is it acceptable to use material left-over from surgical operations for research in general?
- 7.6 Is it acceptable to use material left-over from surgical operations in genetic research? Should there be a different approach for anonymised and for identifiable material?
- 7.7 What types of information do patients need concerning the potential use in medical research of tissue removed during an operation?
- 7.8 Should unexpected findings from genetic research be fed back to the donors of the sample:
 - (1) if the sample was given specifically for research;
 - (2) if the sample was left-over tissue from an operation;
 - (3) only if the person had given consent to such feedback; and
 - (4) if the findings could enable the person to take action to prevent damage to their health, even if the person had not asked to receive this information?

8. Making commercial use of personal genetic information: issues of ownership and property

- 8.1 Concern over some potential commercial uses of genetic information has been expressed both at a personal and a more general level. Some concerns focus on the possibility of commercial exploitation of a person's DNA without the knowledge or consent of that person. Other concerns focus on the potential ethical implications of patenting and the widespread belief that patents will allow for the "commodification" of human genes as they exist in the body. The issue of the patenting has been the subject of intense political debate, but it is now clear that in UK law, human genes as they exist in the cells in our bodies cannot be the subject of patent rights. This part of the consultation document addresses the question of the commercial use of genetic information obtained from the samples of patients and volunteers in research projects. Some patenting issues arise here, but these are of limited application.

The patient's interest in samples: general issues

- 8.2 When genetic testing has been carried out on individuals in the context of their own care or treatment, they expect that those results will be kept confidential. The results will be protected both by the common law duty of confidence, and if stored electronically or in health records, will be subject to the *Data Protection Act* 1998. The thought that those results could be used as a source for commercial gain will most probably never have occurred to the person concerned.
- 8.3 However, the duty of confidence is not absolute. It is well recognised in law that when the public interest in disclosure of information exceeds the duty of confidence to the individual concerned, then information can be passed on. Case law on the scope of the duty of confidence in the health context is limited. However, in December 1999 the Court of Appeal considered the issue in the context of *R v Department of Health, ex parte Source Informatics* which concerned a plan for commercial use of fully anonymised information derived from patients' prescriptions. Lord Justice Simon Brown held that the purpose of the duty of confidence was to protect the patient's privacy, but that the patient had no property rights, or rights to control the use of information referring to him so long as the patient's privacy was not put at risk.
- 8.4 It therefore followed that when information was anonymised, a patient would have no right to prevent the disclosure of that information to a company for the purpose of making commercial gains. However, this of course does not mean that the NHS would contemplate making wholesale disclosure of anonymised information concerning patients to commercial companies – but such a constraint operates as a function of policy and of the ethical codes of professionals rather than as a matter of law.
- 8.5 Lord Justice Simon Brown considered that when anonymised information was disclosed, this could not be seen to thwart the will of the patient or undermine his integrity. However, there is limited information about the extent to which the public actually shares that view. Some members of the public may well feel that a person is in some sense wronged if commercial companies use information about his or her genotype; others may take the view that provided that information is anonymised, this is a useful way of supporting the development of new drugs

and therapies. In this latter view, the common good, which is served by the development of new therapies by pharmaceutical companies, can only be advanced by allowing such practices.

Consent and property issues

- 8.6 In the case of genetic information obtained from samples of volunteers in medical research projects, the issue is whether the subject's consent to participate in the research project covers the commercial use of information obtained from samples. This is an aspect of the consent issue discussed at greater length in Section 6. Where anonymised archived samples are used, commercial issues may also arise, and again the matter requires a balancing of public interest with an awareness of a possible objection to the exploitation of samples that might not have been given with a view to commercial gain being made by another.
- 8.7 One way of approaching these issues is to think in terms of moral claims and entitlements. There is, however, a legal dimension to the problem, which expresses itself in terms of the law of consent (addressed above) and the law of property. The property question here is this: who owns the sample and therefore the DNA which it contains?
- 8.8 Although it is a well-established principle of law that there is no property in a whole dead body, the position of tissue separated from the body is not entirely clear. How the law approaches this issue may depend on how the tissue is separated from the body. The simplest case would be where a person capable of giving consent gave a sample of tissue to a commercial company in the full knowledge that the sample was to be used for commercial purposes. In such cases it is possible to avoid debate about property rights in the tissue by making clear to the person whether or not they will be entitled to any return for their donation.
- 8.9 However, international instruments such as the Council of Europe's *Convention on Human Rights and Biomedicine* (1997) specify that "the human body and its parts shall not, as such, give rise to financial gain". Where processes of work and skill have been applied to the tissue so that it has a value which it would not have had before that work was undertaken, it is permissible to make a profit. As the donor of the tissue would not him or herself have taken part in the process of work or skill, it may be suggested that for the person to make commercial gain out of their own tissue would be against these international instruments. The prospect of people selling bodily organs aroused considerable concern in the 1980's after a kidney-selling scandal. This led to the passage of the *Human Organ Transplants Act* 1989, which aims to prohibit the sale of human organs. The removal of organs obviously has more potentially serious risks to health than does the removal of a small amount of tissue, such as a blood sample; but is there a difference in principle which would justify allowing people to make profit from their tissues but not their organs?
- 8.10 However it is more common for samples to be removed in the context of a person's treatment, for example a surgical operation. Once that tissue has been removed, the ownership of that tissue is a much debated area of law. If the patient consented to the operation on the basis that the removed tissue (such as a gallstone) would be returned to him or her, then (unless returning the tissue would create a public health hazard) it would seem clear that the patient has the right to exclude others from using the tissue.

- 8.11 Few patients make such a request, and samples of tissues are often stored. Some have suggested that the legal position is that either the patient has abandoned the tissue or has made a gift of it to the institution in which it is stored. However, where there is no evidence that the patient intended either result (usually because the situation has never been considered by the patient) the validity of using either analysis is very questionable. Further, when the operation was carried out in the best interests of a person incapable of giving consent, who might never have been capable of forming the intent concerned, the validity of assuming any such intent is even more questionable.
- 8.12 Another analysis suggests that no-one has any rights over the tissue at the moment of its removal but that the person who first acquires it establishes such rights. In the case of surgical operations this would be the institution concerned. Although this could give the institution rights over the tissue, for example to incinerate it (if it was no longer required as part of the clinical record), this does not necessarily mean that the NHS institution could sell the tissue for profit, for the same reasons that individuals may not be able to sell their tissues for profit noted above.
- 8.13 However, this does not exclude the possibility of the tissue being used to make a profit, for example by using it to generate genetic information, which in turn could be the basis of commercial gain. If it were clear that the tissue was the property of the institution then the legal permissibility of such research would be clear. Equally, it would be possible for the NHS organisation to pass it on to a commercial company for such purposes.
- 8.14 Commercial gain very often relies on the patenting of results obtained from research on the sample. Patenting is a method of protecting the results of research so that individuals and companies make the necessary investment to carry out the research. In many areas a major method of knowledge sharing is by the use of patents. Patents provide other researchers with the background to continue experimental work or move into new areas. Without patents, some research might never be carried out because the costs involved could never be recovered. Patents also allow the general public to be informed of the current state of research and its direction and to be involved in debate on ethical issues.
- 8.15 Under patent law the donor, or source of the tissue, would not be entitled to own the patent. However, in the European Directive (98/44/EC) on the legal protection of biotechnological inventions, the issue of donor consent to the filing of a patent for a product derived directly from a tissue sample (but not on information generated from such a sample) was raised in an introductory recital to the Directive. However the wording does not make clear whether the consent refers to the original removal of the tissue or the patent application itself. As recitals do not need to be specifically incorporated into national law, many countries, including the UK, have chosen not to implement any such consent requirement into national law. However as a moral principle and for consistency (and commercial prudence) donors should perhaps understand that commercial gain includes the filing of patents in which they will retain no rights.
- 8.16 Many of the ambiguities described above could be avoided if clearer information was given to those from whom tissue is removed about the potential uses of their sample. The MRC have prepared a model consent form which addresses this issue, as have a number of NHS Trusts. The provision of information enables the

patient to make an informed choice. Mechanisms need to be in place to record and act on conditions placed by the patient, for example that a sample could be used for some purposes but not others. In the National Plan for the NHS, issued in July 2000, the Government indicated that an initiative to improve practice in obtaining consent would be undertaken over the following year. HGC believes that it would be valuable if that initiative took account of the issues arising from the potential uses of human tissue.

- 8.17 The conditions under which tissue can be removed from the dead differ from the conditions under which it can be removed from the living. The law on this subject has been well reviewed in the interim report of the Bristol Inquiry, under the chairmanship of Ian Kennedy. The Court of Appeal has held that there is no property in either a dead body or its parts once they have been lawfully removed from the dead body. However, when a process of work or skill has been applied to parts of the body, property rights may apply and accrue to the person who has applied the process of work and skill. This would suggest that it would be possible to financially gain from such work.
- 8.18 However, when lack of objection from relatives is sought under the *Human Tissue Act* 1961 for lawful removal of tissue from a body for the purposes of research, arguably if the relatives are not told about the potential for using the tissue for commercial gain, and it is subsequently so used, their lack of objection was based on inadequate information. This might be regarded as morally questionable.
- 8.19 In relation to the European Directive on the legal protection of biotechnological inventions referred to above (or in any other circumstance), it is obviously not possible for a dead person to give consent either to the removal of the tissue or to a patent application. It is not clear how the principle of the recital should apply in such circumstances.

Questions: Making commercial use of personal genetic information

- 8.1 Is this a fair representation of the commercial issues? Is there anything else we should have added?**
- 8.2 In what circumstances, if any, should the genetic information of NHS patients be made available to commercial companies engaged in medical research? Should bodily samples taken by the NHS for diagnosis or as a result of surgery be used for commercial research?**
- 8.3 Should specific consent to the use of genetic information for the purposes of commercially-driven medical research be necessary before information is used for such purposes?**
- 8.4 Should the person who is the source of a tissue sample which is detached from his or her body in the course of medical treatment have any rights over what is done with that sample or with the DNA which it contains? If so, what should these rights be?**
- 8.5 The HGC will be monitoring the area of human genetic databases in the future. Are there any particular issues you would like to draw to the Commission's attention?**

9. Protecting the confidentiality of personal genetic information: insurance and employment

Insurance

This consultation document seeks to address some of the issues in relation to disclosure of personal genetic information to insurers. HGC has been asked to give priority to the wider social and ethical issues surrounding genetic tests and insurance. Separately HGC will be taking more detailed evidence from a range of interests in order to review the developments in this area in the three years since the Human Genetics Advisory Commission report “*Implications of Genetic Testing for Insurance*” (available at www.dti.gov.uk/hgac).

The issues

- 9.1 The potential use by insurance companies of genetic information has been the source of some public concern. There is some anxiety that insurance companies may unfairly discriminate against those who, on the basis of the results of genetic tests, are perceived to be at greater risk of developing certain conditions. If this were to become widespread, there could arise a group of people who would find it impossible to obtain certain forms of insurance cover as a result of their having an abnormal genetic test result.
- 9.2 Although such anxieties are an understandable consequence of a scientific development which has attracted so much attention and for which some extreme claims have been made, any response to this issue must be informed by facts. The insurance industry already uses genetic information in the form of family histories.
- 9.3 There is some evidence of unjustifiable genetically-based discrimination in insurance, particularly in the United States. In one study, the relatives of persons with a genetically transmissible disease claimed to have been denied insurance cover even when it had been established that they themselves were not affected by the condition. This suggests that some insurance companies may be tempted to adopt sweeping exclusionary practices if not prevented from doing so. It would not be correct, however, to say that there is current evidence of this happening in the United Kingdom. Indeed, the Code of Practice of the Association of British Insurers (ABI) demonstrates a clear rejection of such an approach. It might also be borne in mind that the interest of insurers in adopting such an approach might be less than might be imagined. Although advances in genetics have made possible certain advances in our ability to predict the development of disease, there is a great amount of disease that cannot be predicted through genetic tests. It is only rare inherited single gene diseases that are likely to be affected in the short term. Other factors, environmental and behavioural, may be more powerful predictors of life expectancy and health in many cases. However, as there is an increase in the range of conditions for which genetic testing becomes possible, the effect of these tests on insurance decisions may grow.
- 9.4 The importance of ensuring fair access to insurance is most pressing in countries where health insurance is required in order to obtain a reasonable level of health care. This is not the case in the United Kingdom (see box). Life insurance is important,

though, as it constitutes an important element in the planning people make for the financial security of their dependants. Although life insurance may commonly be offered with a mortgage for the purchase of a house, it is by no means essential. For these reasons, it is important to ensure that there is no unfair discrimination against certain groups of people who might in this way be deprived of access to a common social good.

Use of genetic information in insurance – UK practice

1997: The Human Genetics Advisory Commission's report – "The Implications of Genetic Testing for Insurance" did not consider that a permanent ban on the use of genetic tests in insurance would be appropriate. However it concluded that "a requirement to disclose results of specific genetic tests... would only be acceptable when a quantifiable association between a given pattern of test results and events actuarially relevant for a specific insurance product had been established". It recommended that the insurance industry should respect a moratorium on requiring disclosure of genetic test results pending review of the actuarial evidence.

October 1998: The Government response to the HGAC report concurred with the Commission that a permanent ban on the use by insurers of genetic information would not be appropriate. It welcomed the ABI Code of Practice (see below).

The Government also established an advisory committee, the Genetics and Insurance Committee (GAIC), which includes representatives of the insurance industry as well as independent geneticist, actuaries and consumer representatives. It has the task of assessing the scientific and actuarial reliability and predictive value of genetic tests.

GAIC has published criteria covering the details of the genetic condition being tested for, the accuracy and reliability of the tests used and the relevance of the test results to decisions about insurance underwriting. GAIC expects that applications will be for genetic conditions caused by changes in a single gene, that are very likely to lead to serious ill health or disability and that are therefore most relevant to the setting of premiums for life and health insurance. GAIC has recently considered an application to use genetic test results for Huntington's Disease in life insurance. It expects to consider further applications relating to Huntington's Disease as well as early-onset forms of Alzheimer's disease and rare inherited forms of breast, colon and other cancers. The intention is to complete a review of these applications by June 2001. GAIC is also working with HGC in the wider review of genetic testing and insurance.

The ABI Code of Practice on Genetic Testing lays down proper practice in relation to the use of genetic test results by insurers. The Code clearly states that there will be no request for a genetic test by any applicant as a precondition of obtaining life insurance, but applicants will be asked to reveal the results of previous tests. This will not be required of applicants for life insurance required to be taken out to obtain a private house purchase mortgage of up to £100,000. The Code also sets out arrangements for appeals against insurers' decisions.

- 9.5 The current UK position represents a compromise. The fact that applicants are not required to undergo testing as a prerequisite means that genetic testing remains fully voluntary. Moreover, while previously obtained test results may be taken into account, this will only be done if the test has been approved by the GAIC. It is

hoped that in this way insurers will give the appropriate weight to genetic factors rather than giving them an exaggerated importance. If insurance companies have access to test results, this also allows them to offer insurance at normal rates to people who have a family history of a genetic condition, but whose test results are normal. Therefore allowing insurers to see genetic test results can work in favour of some applicants.

The arguments

- 9.6 Insurance is a contract between the insurer and the insured, in which the insurer agrees to underwrite a risk in return for a payment from the insured. Some forms of insurance are state-sponsored and are not voluntary agreements between notional equals (National Insurance being an example). For our current purpose, we are concerned with contracts which are freely entered into between the parties, both of whom consider the terms and conditions before entering upon the agreement. The insurance company must assess the risk and decide whether or not to insure it, while the insured person considers the premium and the attached conditions and decides whether or not to accept the terms offered.
- 9.7 In order to assess risk, the insurance company requires all the relevant information it can obtain. This enables an actuarial assessment to be made and an informed commercial decision to be reached. An insurer is not bound to accept a risk which is regarded as unacceptably high, as this would represent a bad bargain from the insurance point of view. Other policy holders who did not represent such a risk might resent paying increased premiums as a result. The majority of life insurance is provided without any information being required from a clinician (it is usually accepted on the basis of a simple statement from the applicant). It may involve a medical examination commissioned by the insurer or a report based on the medical records of the applicant for insurance. The applicant consents to this disclosure of otherwise confidential material to a medical officer of the insurance company. An applicant may also be asked to disclose details of his or her private conduct, including alcohol and tobacco consumption, and details of family background, such as the age at which parents died and the cause of their death. All of this private information is used by the insurer to assess the likely future health and life expectancy of the applicant. A summary of the insurance underwriting process has been prepared by the GAIC and is available at its website (www.doh.gov/genetics/gaic).
- 9.8 Insurers have argued that genetic information is no different from any other medical information in this context, and that they should be allowed to use the results of genetic tests in order to assess the nature of the risk which they are being asked to underwrite. In this view, to exclude genetic information would be arbitrarily to single out one source of information while allowing access to information which is of similar, or even greater predictive value. A simple question as to the cause of parental death may be of far greater predictive value than the result of a genetic test which discloses, say, a 15% greater risk of developing a fatal condition. Allowing insurers to access results of genetic tests can also benefit those people with a family history of a genetic condition, but with a normal genetic test result, to obtain insurance at standard rates.
- 9.9 There are various arguments against the use by insurers of genetic test results. One of these is that genetic testing is so sensitive a matter that it should, as a matter of pure social policy, be kept out of insurance decisions. Genetic testing

is seen as more intrusive than other forms of enquiry or examination, revealing the very “essence” of a person. In this view, access to such information should be denied when the purpose of the access is a commercial or employment decision. This argument is clearly based on notions of privacy, and seeks to limit the occasions for intrusions on this inner sphere. In a more particular sense, a requirement of genetic testing for insurance would mean that a form of testing that is normally freely sought might become less than fully voluntary. There is evidence that many people are unwilling to submit to genetic testing, even when they are aware of a risk which might be confirmed or excluded. If they were required to disclose previous test results or to take genetic tests before obtaining insurance then they might avoid genetic testing completely and lose out on any resulting medical benefits. There is also the question of the right not to know. If in future a genetic test was unwelcome, but necessary to obtain a desired form of insurance, it could impose on people information that they might otherwise not have wanted.

Use of genetic information in insurance – international practice

There is a general consensus in different countries that it is inappropriate to permit insurers to require applicants to undergo genetic testing as a condition of obtaining insurance. This is variously ensured by legislation, voluntary codes and agreements.

The uses to which existing genetic information can be put by insurers in different countries are much more varied. The Netherlands and Sweden have both adopted the approach of setting an ‘enquiry limit’, beyond which the restrictions on the insurers’ use of genetic information do not apply. Others, for example Australia, have rejected this approach.

Additionally in the Netherlands, there is a separate provision that explicitly prohibits the use in insurance of presymptomatic or susceptibility testing for serious, untreatable disorders.

Other countries have voluntary agreements (eg the Swedish State has signed an agreement with insurers governing their use of genetic information) or are currently considering the issues (eg Germany).

Different levels of provision of national health care in different countries means that the focus on particular types of insurance differs in those countries. For example, since the overwhelming majority of Americans rely on health insurance to meet their health care needs, American legislation focuses predominantly on prohibiting unfair discrimination with respect to group health insurance. However, in Australia, the universal provision of health care under the Medicare scheme means that the discrimination issues are of most relevance with respect to life, disability and employment insurance.

Article 11 of the Council of Europe’s *Convention on Human Rights and Biomedicine* (1997) prohibits “any form of discrimination against a person on grounds of his or her genetic heritage”. Article 12 states genetic testing may only be carried out for purposes of health care or research. Exceptions can only be made on very limited public interest grounds set out in Article 26.1 of the Convention. The implications of these provisions for insurance are uncertain.

The European Commission have also recently conducted a study of the regulation of the use of genetic information by insurance companies throughout the EU.

A more detailed comparison of international legislation can be found on the HGC website (www.hgc.gov.uk/business_publications.htm).

- 9.10 A second argument is that genetic risk should be treated simply as “background” risk shared by all and that an assumption of equality should be made. If a person, through no fault of their own, is at a greater genetic risk of developing a condition, then that risk, and the cost it represents, should be distributed in society in general, in the same way as society accepts that some people are going to make much greater demands on health resources than others, but declines nonetheless to discriminate between them in terms of contribution expected. This argument, of course, assumes that insurance is a public matter, to which considerations of community interest might be applied.

- 9.11 A further argument against the use of genetic tests in insurance is that tests will be incorrectly interpreted and given a predictive value that they do not merit. This would lead to unjustifiable decisions to exclude from the insurance pool those whose exclusion could not be defended on actuarial grounds.

Questions: Personal genetic information and insurance

- 9.1 Is this a fair representation of the insurance issues? Is there anything else we should have added?**
- 9.2 Should insurance companies be required to consider personal genetic information differently from other medical information or family history. If so, why?**
- 9.3 In the light of the above questions, what principles should govern the way insurance companies may or may not use pre-existing personal genetic information?**
- 9.4 Should any such principles draw distinctions between:**
- (1) different types of insurance (eg life/health)?**
 - (2) different types of condition (treatable/untreatable)?**
 - (3) the value of the policy to be insured?**
- 9.5 The HGC will be separately considering other aspects of the use of genetic test results in insurance. Are there any particular issues you would like to draw to the Commission's attention?**

Employment

The issues

- 9.12 As in the case of insurance, there has been some public anxiety that employers may use personal genetic information to discriminate improperly against employees who are seen to be at risk of a particular illness or condition. There is no evidence that this being done in the United Kingdom, and indeed when the issue was examined by the HGAC in 1990, only one employer was found to be using genetic testing. This was the Ministry of Defence, which screened applicants for aircrew training for sickle cell disease and trait on the grounds that this condition could cause difficulties when a person so affected is at a high altitude. There is no evidence that the picture disclosed by the Committee has changed for the worse; indeed the MOD is currently reviewing its policy on sickle cell testing.

- 9.13 Employers might in the future wish to engage in genetic testing of employees to identify:
- (1) which employees may be at risk of putting other people at risk in the workplace;
 - (2) which employees have an increased susceptibility to occupational disease; and
 - (3) which employees are likely, because of genetic factors, to experience long, and possibly expensive, periods of absence from work.
- 9.14 A case could be made out for allowing employers to use genetic testing for the first two of these purposes, but there is a clear risk of unfair discrimination if testing for purpose (3) were to become common.

Use of genetic information by employers – UK situation

There is no current legislative prohibition on the use of genetic information in employment. An employer may require an applicant for employment to take a genetic test, and the employer may discriminate, at least in some cases, on the basis of the result of such a test.

Discrimination on the basis of an existing disability of genetic origin would be prevented by the *Disability Discrimination Act* 1995, but there is currently no specific legislation to prevent discrimination against asymptomatic employees. In some circumstances, the requirement for “fair processing” in the *Data Protection Act* 1998 would amount to a prohibition.

HGAC recommended:

- that employees should not be obliged to take a genetic test for employment purposes, and that the right of an individual not to know the result of any test should be respected.
- that employers should not be permitted to use genetic testing to deny employment to employees who might be shown, as a result of the test, to be at a greater risk of general ill health (unrelated to occupational health risk).
- that an employer should only be entitled to ask for access to existing genetic tests if the test relates to a condition which will directly affect the individual’s ability to do the job in question or the individual’s susceptibility to a particular workplace health risk.

The Data Protection Commissioner is consulting on a draft Code of Practice on the Use of Personal Data in Employer/Employee Relationships, which is available from the Data Protection website (wood.ccta.gov.uk/dpr/dpdoc.nsf).

Use of genetic information by employers – practice in other countries

Several European countries have adopted legislation which regulates genetic testing by employers. In Austria, the *Gene Technology Act* 1995 prohibits employers from requesting, collecting or using information derived from genetic tests. Similarly, in Norway and France genetic testing for employment purposes is illegal.

In other countries, such as the Netherlands, Spain and Denmark, genetic tests can only be used by employers where there is an unambiguous health requirement for the job, or where the protection of the employee's health in the workplace calls for such a test.

In the United States, there is evidence of genetic discrimination in employment. Concern over this has resulted in protective legislation in some US jurisdictions and executive action to protect Federal employees against genetic discrimination. The situation in the United States is affected by the desire of some employers to identify potentially unhealthy employees in order to reduce the health insurance commitment that an employer may have to shoulder.

9.15 The Government response to the HGAC *Report on Genetic Testing and Employment* took the form of a letter from Ministers to HGC in July 2000. It invited the HGC to work with other bodies and Government Departments to address two of the main recommendations of the report. These were:

- HGAC's proposed policy principles (see box) which should be observed if and when genetic testing in employment becomes a real possibility; and
- a further review of genetic testing in employment in 5 years time.

The basis for these general principles is covered elsewhere in this document.

HGAC's principles for genetic testing in employment

The HGAC report *The Implications of Genetic Testing for Employment* (1999) proposed a common set of policy principles on genetic testing in employment:

- (1) an individual should not be required to take a genetic test for employment purposes – an individual's "right not to know" their genetic constitution should be upheld;
- (2) an individual should not be required to disclose the results of a previous genetic test unless there is clear evidence that the information it provides is needed to assess either current ability to perform a job safely or susceptibility to harm from doing a certain job;
- (3) employers should offer a genetic test (where available) if it is known that a specific working environment or practice, while meeting health and safety requirements, might pose specific risks to individuals with particular genetic variations. For certain jobs where issues of public safety arise, an employer should be able to refuse to employ a person who refuses to take a relevant genetic test;
- (4) any genetic test used for employment purposes must be subject to assured levels of accuracy and reliability; ... and,
- (5) if multiple genetic tests were to be performed simultaneously, then each test should meet the standards set out in (2), (3) and (4).

Copies of the HGAC report is available at www.dti.gov.uk/hgac/papers/papergl.htm and Government response is available at www.doh.gov.uk/genetics/hgacgovresp.htm

Questions: Personal genetic information and employment

- 9.5 Is this a fair representation of the employment issues? Is there anything else we should have added?**
- 9.6 Do you have any comments on the proposed principles which should govern the way employers use genetic information?**
- 9.7 The HGC will be monitoring the area of genetic information and employment in the future. Are there any particular issues you would like to draw to the Commission's attention?**

10. Personal genetic information in forensic databases

- 10.1 The ability to analyse DNA and the resultant development of national forensic DNA databases is widely recognised as being the most significant breakthrough in crime detection since the introduction of fingerprint identification. Forensic DNA databases provide a similar function to fingerprint indices and permit the comparison of genetic profiles recovered from crime scenes (or taken from suspects and volunteers) with those taken from individuals charged with an offence.
- 10.2 There are however some important limitations to the fingerprint analogy:
- (1) unlike fingerprints, genetic information is shared with biological relatives;
 - (2) DNA is a more reliable informant, being durable, amplifiable from minute samples and recoverable from any human tissue; and
 - (3) DNA is far more information rich than fingerprints. But the DNA profile can only be used for the identification of individuals and will not (as far as is known) provide any information about genetic disorders or susceptibilities. The profile data can however be used to check paternity.
- 10.3 There is considerable public support for the forensic application of genetic information and the UK National DNA Database is set to continue to develop and grow significantly. But some concerns have also been expressed about certain aspects of the existing arrangements and the direction of future developments. These concerns centre on: the scope of police sampling policy; the management of the National DNA Database; and issues of consent.

Legal basis for forensic collection and use

- 10.4 In 1984, the *Police and Criminal Evidence Act* (PACE) limited the taking of bodily samples to people suspected of “serious arrestable offences” and where it was relevant to the investigation of this offence. There is no absolute definition of these type of offences but they would include: serious crimes against the person (eg murder and rape); threats which if carried out would be likely to lead to serious interference with the administration of justice; or substantial financial gain or serious financial loss to any person. The development of DNA profiling prompted the amendment of PACE by the *Criminal Justice and Public Order Act* (CJ&PO) 1994. This extended the circumstances in which bodily samples could be taken and made it possible to operate an effective National DNA Database. These amendments to PACE also introduced safeguards relating to the circumstances in which DNA samples could be taken, used and destroyed. In Scotland, the relevant legislation, which has comparable provisions, is the *Criminal Procedure (Scotland) Act* 1995.
- 10.5 In England and Wales, bodily samples may now be taken in broadly the same circumstances as fingerprints. Non-intimate samples (mouth swabs and hair) can be taken, without consent, from any person suspected of being involved in, charged with or about to be reported for, or convicted of a “recordable offence”. Recordable offences include the majority of offences which the police investigate, including those which involve violence or dishonesty and that can lead to a prison sentence.

- 10.6 Intimate samples may be taken provided the appropriate consent is given. The police also have the power under PACE to seize a dead body or take a sample from it if there is reasonable suspicion that the deceased person may have committed an offence. Individuals may also voluntarily participate in police mass screening exercises by providing DNA samples.
- 10.7 If a person is not suspected of having committed an offence, is not prosecuted or is acquitted, then the DNA sample that they provided must be destroyed after the conclusion of the due legal process. Samples need not be destroyed if another person from whom a DNA sample was taken as part of the same investigation is convicted of the offence, to allow for further analysis if the case is reviewed. But for both types of sample the resulting information cannot be used in evidence against the provider or for any investigation of an offence. It is important to note that although samples are destroyed when the suspect is not prosecuted or is acquitted, the DNA profile derived from that sample remains on the database. This has been justified on the grounds that this assisted to build up statistical information on the incidence of the same profile occurring in unrelated persons. It is questionable whether this justification still applies, and a question might be raised as to why the continued retention of these profiles takes place, even if their use in criminal investigation is currently suppressed. These samples will add considerably to the size of the database.
- 10.8 In July 1999, the Home Office issued “Proposals for Revising Legislative Measures on Fingerprints, Footprints and DNA Samples”. The aim, in the case of DNA samples, was to review the legislation in the light of experience and the need to reflect new practices and procedures. There were three proposals relating to DNA samples. These were:
- i. That DNA samples, and the information derived from them, should be retained and used in future investigations with a volunteer’s written consent (which they would have the right to subsequently withdraw). This addressed police concern that an increasing number of people would refuse to co-operate when approached repeatedly to act as volunteer donors in mass screening exercises, because the law required that their previous sample, and the associated DNA profile, be destroyed.
 - ii. That DNA samples should be retaken where a full DNA profile cannot be produced because of scientific failure or inadvertent or malicious destruction of the sample prior to analysis.
 - iii. That statutory powers should be made explicit to enable DNA samples taken here, and the information derived from them, to be compared with those from other jurisdictions (ie the linking of national DNA databases).

The seriousness of offences for which DNA samples are taken

- 10.9 Since the inception of the National DNA Database in 1995 there has been a trend towards the expansion of the categories of offences for which bodily samples are taken from the original criteria of burglary, violence and sex offences. Some forces now include drug and vehicle related crimes and in the Lothian region of Scotland people stopped for moving traffic offences can be asked for a DNA sample. In the latter situation one could question the relevance of taking a DNA sample to this form of motor offence, unless the individual was also suspected of

committing a more serious offence. Overall, the scope for sample collection under UK law, the growing number of situations in which bodily samples are being taken and the recent proposals to seek consent to retain more samples, could lead to the suspicion that a comprehensive DNA database is being built up by stealth. Cost is also a factor here. In the early days of the National DNA database, the cost of taking and analysing samples was a barrier to the processing of profiles. This provided an element of selection based on the seriousness of the matter under investigation and the relevance of a DNA profile to its conclusion. The lowering of the cost base has meant that many more profiles are now being generated than may have been originally envisaged.

Police access to health care records containing genetic information

- 10.10 A distinction should be drawn between allowing police direct access to health care records, and disclosing information contained in health care records to the police. As a matter of practice, police direct access to records, or copies of records, would normally require a relevant court order. Information contained in records may be disclosed to the police with the consent of the patient.
- 10.11 If, for whatever reason (eg possible increased danger to others), consent to disclosure is not sought or is refused, information from records may be passed to the police if the public interest in disclosure exceeds the duty of confidence to the patient. In Department of Health guidance (HSG(96)18) it was considered that it might be justifiable to pass on information if the following conditions were satisfied:
- i. without disclosure, the task of preventing, detecting or prosecuting a serious crime would be seriously prejudiced or delayed;
 - ii. information is limited to what is strictly relevant to a specific investigation; and
 - iii. there are satisfactory undertakings that the information will not be passed on or used for any purpose other than the investigation in hand.
- 10.12 The guidance also stated that requests for information relating to a number of patients in order to identify one or more ("fishing expeditions") is likely to be justified only if there is a very strong public interest. In practice, it will be extremely rare for such disclosure to be justifiable.
- 10.13 In practice, it is quite rare for disclosure to be sought from medical practitioners (as distinct from providing reports on defendants).

The National DNA Database and its management

- 10.14 The National DNA Database is operated by the Forensic Science Service (FSS), who primarily provide a national service to United Kingdom police forces, the Crown Prosecution Service, Customs and Excise and other agencies. The Database is an investigative tool which the police and other authorised agencies are not obliged to use. Samples are analysed to generate DNA profiles which are put on the database and checked against other profiles obtained from scenes of crime. This information is held and used in accordance with the requirements of the 1998 *Data Protection Act*.

- 10.15 The FSS also provide a corporate service. A Company DNA database is compiled containing the DNA profiles of personnel and possibly family members to assist in the speedy authentication of extortion claims in a kidnap situation. The information associated with each sample is agreed with the customer and should be sufficient to guarantee its integrity but preserve its anonymity. The profiles may be held by the FSS on a secure stand-alone database or supplied to the customer in written or electronic format.
- 10.16 National forensic DNA databases are growing in number and size. UK law, allowing the collection of DNA samples from suspects and, with consent, from the general public in mass screening exercises, gives more scope for the inclusion of individuals than is the case in most other countries. The UK database was the first of its kind in the world and it is the most comprehensive, with around 750,000 samples rising at a rate of about 6,000 a week and over 70,000 identifications. The Government is committed to its further development and it is projected that the samples and profiles of one third of the UK male population will eventually be on the database.

International comparisons

The UK, France, Germany, the Netherlands, Austria, Switzerland, the US, Canada and Australia all operate forensic DNA databases of similar construction and a number of European bodies are concerned with co-operation and standardisation.

In addition to the UK, Australia, Canada and Germany have conducted mass screening exercises, but this is not the case in the US where Fourth Amendment protections would make this difficult without reasonable suspicion.

In France samples may only be collected by the police from convicted sexual offenders and must be destroyed after 40 years or whenever an offender reaches the age of 80.

In the US samples could also originally only be taken from convicted sexual offenders. But this was soon extended to other violent offences including murder and crimes against children and the trend is increasingly towards the inclusion of all felonies (eg forgery and burglary) and of certain types of juvenile offenders. Some US authorities are now also beginning to call for a similar system to that in the UK where samples may be taken from suspects. Although if arrest does not lead to conviction samples are usually destroyed, in many US States the wrongly convicted must petition to have their DNA records removed.

In Canada samples are collected from all those convicted of serious violent offences but for serious non-violent offences such as robbery, this is left to the discretion of magistrates.

Consent and Confidentiality

- 10.17 The same degree of consent is clearly not required for at least some forensic uses of genetic information as it is in the medical field. Legislation and international conventions and instruments specifically differentiate between forensic and medical use. As has been said, non-intimate samples can be taken, without consent, from those suspected of committing recordable offences.
- 10.18 Where consent is required, police forces appear to have considerable autonomy in how this is obtained. There appears to be no systematic approach and it is not clear whether or not written consent is always obtained and whether this is supported by the provision of written information.
- 10.19 In situations where individuals are asked to voluntarily consent to participate in police mass screening exercises, the fact that failure to consent could invoke police suspicion challenges the notion that consent is truly voluntary.
- 10.20 In some circumstances bodily samples are lawfully retained for future possible reprofiling. Any other use of identifiable samples, for example for forensic or medical research, would normally require specific consent and the associated information would still be subject to the *Data Protection Act* 1998. At present Home Office guidance links retention of the profile to the retention of the criminal record on the police national computer. These retention periods are based on agreed criteria and are subject to change. It would be important to avoid a situation where the core criminal record (including fingerprints) has been deleted but the DNA profile is retained. Recent reports have criticised the quality of the records and failure to delete records in a timely manner. The linkage of deletion of DNA profiles to these records raises obvious concerns.

Questions: Personal genetic information in forensic databases

- 10.1 **Is this a fair representation of the forensic issues? Is there anything else we should have added?**
- 10.2 **For what type of criminal offence, if any, should it be allowable to include a person's bodily sample and DNA profile on a forensic database?**
- 10.3 **Should a person's bodily sample and/or DNA profile remain on the database indefinitely once he or she has been convicted of a relevant criminal offence, or should continued inclusion be subject to review?**
- 10.4 **In what circumstances, if any, should genetic information from a person's medical records be given to the police?**
- 10.5 **Should third party researchers have access to a forensic DNA database for research purposes?**
- 10.6 **The HGC will be monitoring the area of forensic DNA databases in the future. Are there any other particular issues you would like to draw to the Commission's attention?**

11. Further comments

Other issues

- 11.1 In preparing this discussion document the HGC Working Group has aimed to take a broad look at the different aspects of the storage, protection and use of personal genetic information.

Questions: Other information

- 11.1 Does this discussion document fairly represent the issues associated with the treatment of personal genetic information? Are there any other relevant issues on which you would like to comment?**

Evaluation

- 11.2 We are interested in ensuring that this public consultation has been fairly and effectively run. We would be grateful if you could answer the following evaluation questions by, in each case, ticking one option.

Questions: Evaluation

- 11.2 In my opinion, the consultation has been presented in an unbiased way.**

Agree ☐ Neither agree nor disagree ☐ Disagree ☐

- 11.3 The consultation has taken place at a sufficiently early stage in the policy formation process to allow participants to have some genuine influence.**

Agree ☐ Neither agree nor disagree ☐ Disagree ☐

- 11.4 I have confidence that the overall views of respondents to this consultation will form the basis for HGC's later recommendations.**

Agree ☐ Neither agree nor disagree ☐ Disagree ☐

- 11.5 In my opinion, I was provided with sufficient time and background information in order to effectively take part in the consultation.**

Agree ☐ Neither agree nor disagree ☐ Disagree ☐

- 11.6 In my opinion, the nature and scope of the consultation was well defined (ie I understood precisely what was required of me).**

Agree ☐ Neither agree nor disagree ☐ Disagree ☐

Additional information

- 11.3 We would be grateful if you could provide us with your contact details to aid in our analysis of responses and to let us know if you wish to receive further information about this or future HGC consultations.

(BLOCK CAPITALS PLEASE)

Title _____ **Forename/s** _____ **Surname** _____

Organisation (if applicable) _____

Address _____

Postcode _____ **e-mail** _____

I would be happy if my response were made publicly available ☐

I would be interested in future consultations ☐

I would like a summary of the outcome of this consultation ☐

Unless you do not wish to complete the evaluation section or provide your contact details, after completion please remove or copy this page and attach it to the top of your overall response

Annex 1: The Working Group on the Storage, Protection and Use of Genetic Information

Terms of reference

- 1) To develop a set of general principles relating to the storage, protection and use of genetic information for approval by HGC.
- 2) To collate information on the existing protections for the storage and use of personal genetic information across the range of areas in which such information is likely to arise.
- 3) To review the range of purposes/uses for which personal genetic data may be used including research (including databases), clinical, insurance, employment, forensic, public/commercial partnerships etc, taking account of the work of HGC's Horizon-Scanning Sub-group.
- 4) To identify and summarise the relevant protections, review their adequacy in the light of potential uses and highlight any inconsistencies or gaps in those protections, taking account of relevant European initiatives, international agreements and the experience of other countries in these areas.
- 5) To work with the Sub-group on Public Involvement in Genetics to develop a proposed strategy for public and professional involvement in these issues for consideration by HGC.
- 6) To provide regular progress reports to HGC.

Members

Professor Alexander McCall Smith (Chair, HGC Member)
Professor of Medical Law, University of Edinburgh

Dr Elaine Gadd (Co-opted Member)
Senior Medical Officer, Medical Ethics, Department of Health

Professor John Harris (HGC Member)
Sir David Alliance Professor of Bioethics, University of Manchester

Professor Norman Nevin (HGC Northern Ireland CMO Representative)
Professor of Clinical Genetics at Belfast City Hospital

Ms Hilary Newiss (HGC Member)
Solicitor

Reverend John Polkinghorne (HGC Member)
Canon Theologian of Liverpool; formerly President of Queen's College Cambridge

Professor Martin Richards (HGC Member)
Professor of Family Research, Centre for Family Research, University of Cambridge

Annex 2: Members of the Human Genetics Commission

Chair

Baroness Helena Kennedy QC
(barrister and broadcaster)

Vice-Chair

Professor Alexander McCall Smith
(Professor of Medical Law, University of Edinburgh)

Members

Dr William Albert
(Chair of the International Committee of the British Council of Disabled People)

Professor Elizabeth Anionwu
(Professor of Nursing, Head of Mary Seacole Centre for Nursing Practice)

Professor John Burn
(Professor of Clinical Genetics, University of Newcastle upon Tyne and Director, Northern Genetics Service)

Professor John Durant (until October 2000)
(Head of Science and Communication at the Science Museum, London and Professor of Public Understanding, University of London)

Ms Ruth Evans
(formerly Director of the National Consumer Council)

Professor Peter Goodfellow
(Senior Vice-President, Discovery Worldwide, SmithKline Beecham Pharmaceuticals)

Dr Hilary Harris
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Dr Gillian Samuels
(Director of Science Policy (Europe), Pfizer)

Mr Geoffrey Watts
(journalist and presenter of BBC Radio 4's Leading Edge)

Mr Philip Webb
(Co-optee on Board of Trustees of Genetic Interest Group)

Professor Veronica van Heyningen
(Head of Cell Genetics Section, MRC Human Genetics Unit, Edinburgh)

Ex Officio Member

Ms Ruth Deech
(Chair of Human Fertilisation and Embryology Authority)

Representatives of the Chief Medical Officers

Each of the four UK Chief Medical Officers will be able to participate in HGC or nominate a representative with observer status.

Mrs Jackie Axelby (England)
(Chief Executive, Northumberland Health Authority)

Professor Peter Harper (Wales) (from September 2000)
(Professor and consultant in Medical Genetics, University of Wales)

Dr Patrick Morrison (Northern Ireland) (from December 2000)
(Consultant Clinical Geneticist, Belfast City Hospital)

Professor Norman Nevin (Northern Ireland) (until December 2000)
(Professor of Clinical Genetics at Belfast City Hospital)

Dr Rosalind Skinner (Scotland)
(Principal Medical Officer of Public Health Medical Division, SEHD)

Annex 3: House of Lords Inquiry into human genetic databases

The House of Lords Select Committee on Science and Technology is investigating current and planned human genetic databases. In July 2000 it invited written evidence on the following questions:

- (1) What current projects involve collecting genetic information on people in the UK? What other projects are about to start? Are there collections of material (eg tissue samples) that could be used to generate databases of DNA profiles?
- (2) Why are these genetic databases being assembled? How are these activities funded? What practical considerations will constrain developments? Are there alternative ways of fulfilling the objectives?
- (3) What is the genetic information that is being collected? How is it being stored and protected?
- (4) How do the organisations involved see their responsibilities regarding privacy; consent; future use; public accountability; and intellectual property rights?
- (5) How do they see their activities in the area of genetic databases developing in the future? What advances in sequencing, screening and database technology are they anticipating?
- (6) What lessons should be learnt from genetic database initiatives in other countries?

As noted in paragraph 1.5, the written evidence submitted to the Inquiry was published on 8 November 2000 as *Human Genetic Databases: written evidence* (HL Paper 115, ISBN 0 10 411500 9), the contents of which are also available on the House of Lords website (www.publications.parliament.uk/pa/ld/ldsctech.htm). This will form the basis for the Select Committee's further investigation at a series of public hearings of oral evidence between December 2000 and February 2001. The Committee will produce a report to the House of Lords in March 2001, with recommendations addressed to Government, the HGC (as it begins to finalise its draft guidelines on the storage, protection and use of genetic information) and others.

Questions about the progress of the Inquiry (including the possibility of late submission of written evidence on the questions above) should be addressed to the Clerk of the Inquiry, Roger Morgan – telephone 020 7219 6072 or email morganr@parliament.uk.

